

On Monday, you explored your DNA data. Today, we will take it further and find out what you need to know about your SNPs and their functionality.

Our first example will be *PON1* gene rs662

<https://www.ncbi.nlm.nih.gov/pubmed/>

Free radicals

PON1 rs662 CT/CC THEN tomato >5 g/kg or lycopene >0.5 mg/kg

**rs662 is also known as Q192R**

Br J Nutr. 2005 Mar;93(3):291-7.

**Paraoxonase 1 Q192R (PON1-192) polymorphism is associated with reduced lipid peroxidation in healthy young men on a low-carotenoid diet supplemented with tomato juice.**

Bub A1, Barth SW, Watzl B, Briviba K, Rechkemmer G.

#### **Abstract**

The HDL-bound enzyme paraoxonase (PON) protects LDL from oxidation and may therefore attenuate the development of atherosclerosis. We examined the effect of tomato and carrot juice consumption on PON1 activity and lipid peroxidation in healthy young volunteers with different PON1-192 genotypes (Q/R substitution at position 192). In this randomized cross-over study twenty-two healthy, non-smoking men on a low-carotenoid diet received 330 ml/d tomato juice (37.0 mg lycopene, 1.6 mg beta-carotene) or carrot juice (27.1 mg beta-carotene, 13.1 mg alpha-carotene) for 2 weeks. Intervention periods were preceded by 2-week low-carotenoid intake. We determined the PON1-192 genotype by restriction fragment length polymorphism-polymerase chain reaction (RFLP-PCR) and measured ex vivo LDL oxidation (lag time), plasma malondialdehyde and PON1 activity at the beginning and end of each intervention period. At baseline, lag time was higher ( $P < 0.05$ ) in QQ (111 (sd 9) min) than in QR/RR subjects (101 (sd 8) min). Neither tomato nor carrot juice consumption had significant effects on PON1 activity. However, tomato juice consumption reduced ( $P < 0.05$ ) plasma malondialdehyde in QR/RR (Delta:  $-0.073$  (sd 0.11) micromol/l) as compared to QQ subjects (Delta:  $+0.047$  (sd 0.13) micromol/l). Carrot juice had no significant effect on malondialdehyde irrespective of the PON1-192 genotype. Male volunteers with the QR/RR genotype showed an increased lipid peroxidation at baseline. Although tomato and carrot juice fail to affect PON1 activity, tomato juice intake reduced lipid peroxidation in healthy volunteers carrying the R-allele of the PON1-192 genotype and could thus contribute to CVD risk reduction in these individuals.

Exp Biol Med (Maywood). 2016 Mar 27. pii: 1535370216641786. [Epub ahead of print]

**The common variant Q192R at the paraoxonase 1 (PON1) gene and its activity are responsible for a portion of the altered antioxidant status in type 2 diabetes.**

Zargari M1, Sharafeddin F1, Mahrooz A2, Alizadeh A3, Masoumi P1.

#### **Abstract**

In this study, we investigated the effects of paraoxonase 1 (PON1) activities and the variant PON1-Q192R on the ferric reducing ability of plasma (FRAP) and total thiol. In addition, we examined the distribution of genotypes of this variant and the relationship of the genotypes with age in patients with type 2 diabetes (T2D). A total of 115 patients with T2D were enrolled in this study. Paraoxonase activity (PON-para) and arylesterase activity (PON-aryl) were determined using spectrophotometric assays. The distribution of the Q192R genotypes was determined by the double substrate method. The antioxidant status was evaluated by determining FRAP and total thiol. The frequencies of Q and R allozyme were 0.78 and 0.22, respectively. The multivariate analysis identified a significant association between the variables PON1-Q192R (Wilks'  $\lambda = 0.85, P = 0.002$ ) and PON-aryl (Wilks'  $\lambda = 0.896, P = 0.017$ ), with FRAP and total thiol. The significant difference observed for PON1-Q192R and PON-aryl is primarily due to the changes in FRAP levels ( $\eta^2 = 0.127, P = 0.002$  for PON1-Q192R;  $\eta^2 = 0.083, P = 0.011$  for PON-aryl). The interaction PON1-Q192R-PON-aryl increased the effect sizes from 8 to 19% for FRAP. Only in R-carrying genotypes, there were significant correlations between both PON-para/HDL ( $r = -0.574, P < 0.001$ ) and PON-aryl/HDL ( $r = -0.577, P < 0.001$ ) with age. Our data suggest that the variant PON1-Q192R and PON1 activity, particularly PON-aryl, influenced the antioxidant status in T2D. The interaction of this variant and PON1 activity increased the effect size on the antioxidant capacity. Moreover, the presence of the R allozyme may potentiate the effects of age on susceptibility to cardiovascular diseases in T2D.

## Paraoxonase 1 polymorphisms (L55M and Q192R) as a genetic marker of diabetic nephropathy in youth with type 1 diabetes.

Fekih O1, Triki S, Rejeb J, Neffati F, Douki W, Ommezzine A, Chouchane S, Guediche MN, Bouslama A, Najjar MF.

### Abstract

#### INTRODUCTION:

Paraoxonase 1 (PON1) polymorphisms have been largely involved in diabetes complications. The aim of the study is to evaluate effects of PON1 polymorphisms (L55M and Q192R) on Diabetic nephropathy (DN).

#### MATERIAL AND METHODS:

The study involved 116 children and adolescents with Type 1 diabetes (T1D) and 91 healthy subjects. Albumin excretion rate (AER) was determined by immunoturbidimetry. PON1 activity was measured by a spectrophotometric method and genotyping of PON1 gene was assessed by multiplex PCR followed by RFLP.

#### RESULTS:

PON1 activity was inversely correlated to AER ( $r = -0.245$ ,  $p = 0.008$ ). A significant decrease ( $p = 0.037$ ) in PON1 activity has been shown between patients with nephropathy and those without (162 (57 - 618) Vs 316 (37 - 788) IU/L, respectively). The distribution of AER was, for L55M polymorphism MM > LM > LL ( $p = 0.002$ ) and for Q192R polymorphism QQ > QR > RR ( $p < 0.001$ ). The opposite distribution was noted for PON 1 activity ( $p < 0.001$ ). LMQQ and MMQQ haplotypes seem to increase AER ( $p = 0.004$ ,  $p = 0.003$ , respectively) and to reduce PON1 activity ( $p = 0.011$ ,  $p = 0.052$ , respectively) in youth with T1D. However LLRR haplotype seems to have the opposite effect.

#### CONCLUSION:

This study demonstrated that PON1 polymorphisms L55M and Q192R seem to be a genetic marker involved in the development of DN in T1D.

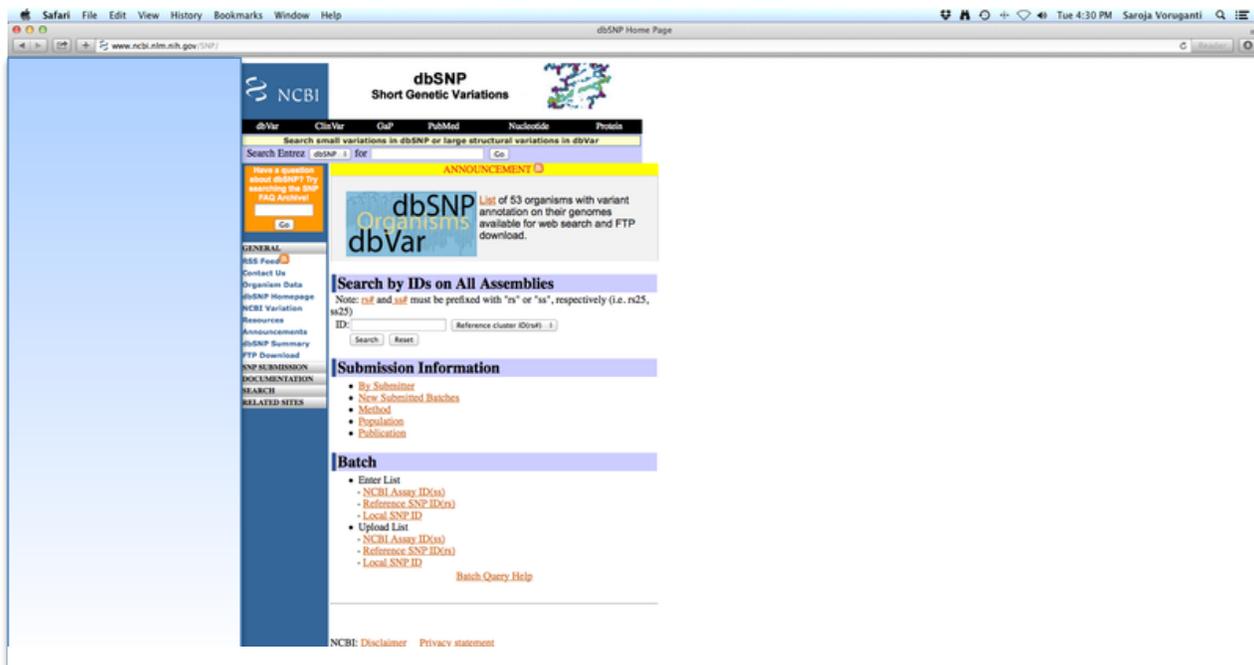
### Workshop Task

dbSNP – NCBI database of genetic variation

1. dbSNP page

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

<http://www.ncbi.nlm.nih.gov/books/NBK44455/>



The screenshot displays the NCBI dbSNP website. At the top, there are navigation tabs for dbVar, ClinVar, GSP, PubMed, Nucleotide, and Proteins. Below these is a search bar with the text "Search small variations in dbSNP or large structural variations in dbVar". The main content area includes an announcement for "dbSNP Organizations dbVar" and a section for "Search by IDs on All Assemblies" with a note that "rs" and "rs" must be prefixed with "rs" or "rs", respectively. There is also a "Submission Information" section with links for "By Submitter", "New Submitted Batches", "Method", "Population", and "Publication". A "Batch" section lists options for "Enter List", "NCBI Assay ID(s)", "Reference SNP ID(s)", "Local SNP ID", "Upload List", "NCBI Assay ID(s)", "Reference SNP ID(s)", and "Local SNP ID".

2. Enter rs662 into the search panel

dbSNP Search Results for rs662

Results: 6

- rs662 (Homo sapiens)
  - Chromosome: 7:95308134
  - Gene: PON1 (GeneView)
  - Functional Consequence: missense
  - Alele Origin: G(gemine)/A(gemine)
  - Clinical significance: other
  - Validated: by 1000G.by cluster.by frequency.by hapmap.by submitter
  - Global MAF: T=0.45712289
  - HQVS: NC\_000007.13:g.94937446T>C, NC\_000007.14:g.95308134T>C, NG\_008779.1:g.21439A>G, NM\_000448.5:c.575A>G, NP\_000437.3:p.Gln192Arg
- rs11567868 has merged into rs662 (Homo sapiens)
  - Chromosome: 7:95308134
  - Gene: PON1 (GeneView)
  - Functional Consequence: missense
  - Alele Origin: G(gemine)/A(gemine)
  - Clinical significance: other
  - Validated: by 1000G.by cluster.by frequency.by hapmap.by submitter
  - Global MAF: T=0.45712289
  - HQVS: NC\_000007.13:g.94937446T>C, NC\_000007.14:g.95308134T>C, NG\_008779.1:g.21439A>G, NM\_000448.5:c.575A>G, NP\_000437.3:p.Gln192Arg
- rs13306997 has merged into rs662 (Homo sapiens)
  - Chromosome: 7:95308134
  - Gene: PON1 (GeneView)
  - Functional Consequence: missense
  - Alele Origin: G(gemine)/A(gemine)
  - Clinical significance: other
  - Validated: by 1000G.by cluster.by frequency.by hapmap.by submitter
  - Global MAF: T=0.45712289
  - HQVS: NC\_000007.13:g.94937446T>C, NC\_000007.14:g.95308134T>C, NG\_008779.1:g.21439A>G, NM\_000448.5:c.575A>G, NP\_000437.3:p.Gln192Arg

3. Click on rs662
4. Information about rs662 provided.

Reference SNP (rs662) Cluster Report: rs662

Organism: human (Homo sapiens)

Molecule Type: Genomic

Created/Updated in build: 36/147

Map to Genome Build: 107/Weight 1

Validation Status: [P](#) [C](#) [H](#)

Citation: [PubMed](#)

Variation Class: SNV: single nucleotide variation

RefSNP Alleles: A/G (REV)

Alele Origin: Argemine/G(gemine)

Ancestral Allele: G

Variation Viewer: [View](#)

Clinical Significance: other

MAF/MinorAlleleCount: C=0.377045735 (EAAC), T=0.45712289 (1000 Genomes), C=0.41229361 (GO-ESP)

HQVS Names:
 

- NC\_000007.13:g.94937446T>C
- NC\_000007.14:g.95308134T>C
- NG\_008779.1:g.21439A>G
- NM\_000448.5:c.575A>G
- NP\_000437.3:p.Gln192Arg

Assembly	Release	Chr	Chr Pos	Contig	Contig Pos	SNP in Chr	Contig allele	Contig in Chr	Neighbor SNP	Map Method
GRCh38.p2	107	Z	95308134	NT_007933.16	3281355	Rev	T	For	site	mapup
GRCh37.p13	105	Z	95321448	NT_007933.15	3281358	Rev	T	For	site	blast

GeneView via analysis of contig annotation: [PON1](#) paroxonase 1

View more variation on this gene (click to hide)

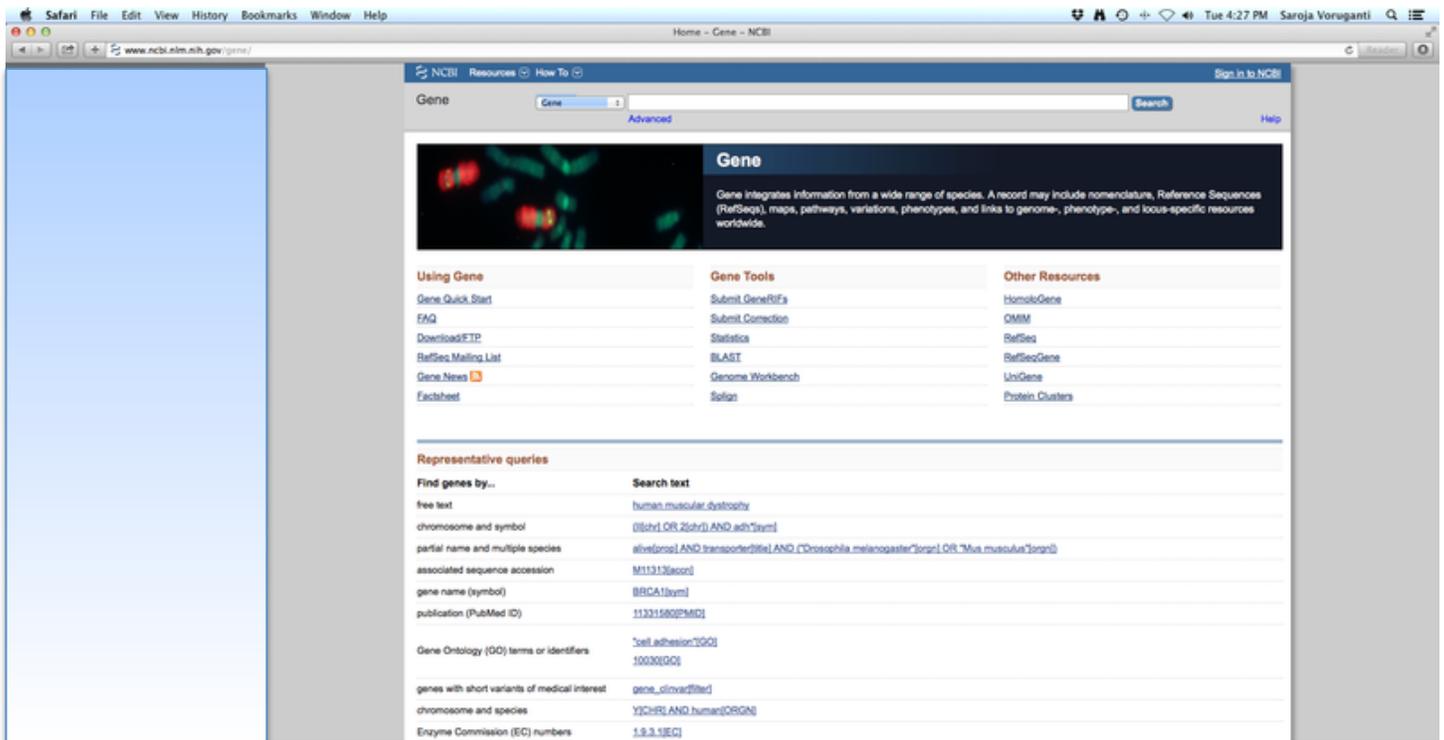
Clinical Source:  in gene region  cSNP  has frequency  double hit [Go](#)

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Alele
GRCh38.p2	Rev	7	95308134	NT_007933.16	3281355	T

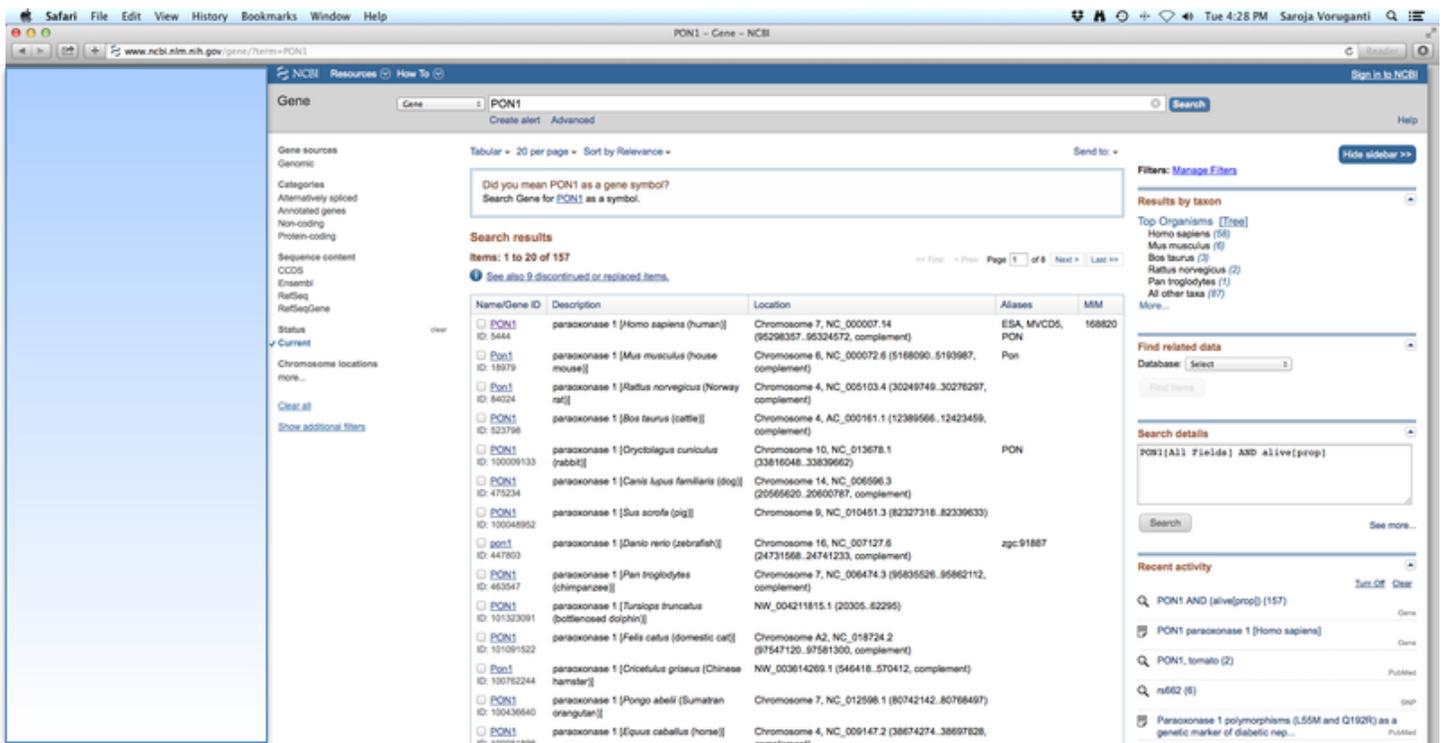
RefSeqGene Mapping	RefSeqGene	Gene (ID)	SNP to RefSeqGene	Position	Alele
NG_008779.1	<a href="#">PON1</a> (5444)	PON1	For	21439	A

Gene Model(s)	Function	SNP to mRNA	Accession	Position	Alele change	Accession	Position	Residue change
missense	For	NM_000448.5	572	C>A	NP_000437.3	192	G [Gln] => R [Arg]	

5. For information on the gene *PON1*  
<http://www.ncbi.nlm.nih.gov/gene/> - A portal to gene-specific content based on NCBI's RefSeq project, information from model organism databases, and links to other resources.



6. Enter the word PON1 in search



7. Click on Paroxanase 1 (Homo Sapiens (human))

**Gene**

Full Report -

**PON1 paraoxonase 1 [Homo sapiens (human)]**  
Gene ID: 5444, updated on 15-May-2016

**Summary**

**Official Symbol:** PON1 provided by tSNC  
**Official Full Name:** paraoxonase 1 provided by tSNC  
**Primary source:** HGNC:HGNC:9204  
**See related:** Ensembl:ENSG000000005421; HPSD:01361; MM:168620  
**Gene type:** protein coding  
**REVIEWED status:** REVIEWED  
**Organism:** *Homo sapiens*  
**Lineage:** Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorhini; Catarrhini; Hominoidea; Homo  
**Also known as:** ESA; PON; MVCD5  
**Summary:** The enzyme encoded by this gene is an arylesterase that mainly hydrolyzes paraoxon to produce p-nitrophenol. Paraoxon is an organophosphorus anticholinesterase compound that is produced in vivo by oxidation of the insecticide parathion. Polymorphisms in this gene are a risk factor in coronary artery disease. The gene is found in a cluster of three related paraoxonase genes at 7q21.3. [provided by RefSeq, Oct 2008]  
**Orthologs:** [mouse](#)

**Genomic context**

Location: 7q21.3 [See PON1 in Genome Data Viewer](#) [Epigenomics Map Viewer](#)  
Exon count: 9

Annotation release	Status	Assembly	Chr	Location
107	current	GRCh38.p2 (GCF_000001495.28)	7	NC_000007.14 (95296357..95324572, complement)
105	previous assembly	GRCh37.p13 (GCF_000001495.25)	7	NC_000007.13 (94927069..94953884, complement)

Chromosome 7 - NC\_000007.14

**Genomic regions, transcripts, and products**

Genomic Sequence: [NC\\_000007.14 Chromosome 7 Reference GRCh38.p2 Primary Assembly](#)

**Table of contents**

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- Genomic context
- Genomic regions, transcripts, and products
- Bibliography
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- Markers, Potential readthrough, Homology, Gene Ontology
- General protein information
- NCBI Reference Sequences (RefSeq)
- Related sequences
- Additional links
- Locus-specific Databases

**Genome Browsers**

- Genome Data Viewer
- Map Viewer
- Variation Viewer (GRCh37.p13)
- Variation Viewer (GRCh38)
- 1000 Genomes Browser (GRCh37.p13)
- Ensembl
- UCSC

**Related information**

- Order cDNA clone
- BioAssay
- BioAssay by Target (List)
- BioAssay by Target (Summary)
- Bookshelf for PON1

8. HapMap <http://hapmap.ncbi.nlm.nih.gov> - The HapMap is a catalog of common genetic variants that occur in human beings. It describes what these variants are, where they occur in our DNA, and how they are distributed among people within populations and among populations in different parts of the world.

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

**Project Information**

- About the Project
- HapMap Publications
- HapMap Tutorial
- HapMap Mailing List
- HapMap Project Participants

**Project Data**

- HapMap Genome Browser release #28 ( Phases 1, 2 & 3 - merged genotypes & frequencies )
- HapMap3 Genome Browser release #3 ( Phase 3 - genotypes & frequencies )
- HapMap Genome Browser release #27 ( Phase 1, 2 & 3 - merged genotypes & frequencies )
- HapMap3 Genome Browser release #2 ( Phase 3 - genotypes, frequencies & LD )
- HapMap Genome Browser release#24 ( Phase 1 & 2 - full dataset )
- GWAs Karyogram
- HapMap
- HapMap FTP
- Bulk Data Download
- Data Freezes for Publication
- ENCODE Project
- Guidelines For Data Use

**Useful Links**

- TSC SNP Downloads
- HapMap Samples at Coriell Institute
- HapMap Project Press Release
- NIH/NIH HapMap Page
- NCBI Variation Database (dbSNP)
- Japanese SNP Database (JSNP)

**News**

- 2013-06-14: HapMap data conversion tool**  
There are several inquires for a conversion tool to convert HapMap data into the VCF format. Please take a look of [The Genome Analysis Toolkit](#) (by Broad Institute).
- 2012-12-06: Downtime for hardware maintenance**  
From December 15 - 16, Hapmap site will be taken offline for an internal hardware maintenance. Sorry for the inconvenience.
- 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 ref 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 release #28 (NCBI build 36, dbSNP b126). Data is available for bulk download and also available for browsing. Click here to read the latest release notes.
- 2010-05-28: HapMap3 Public Release #3**  
Genotypes and frequency data in hapmap format are now available for data in HapMap phase 3 release #3 (NCBI build 36, dbSNP b126). Data is available for bulk download and also available for browsing. Click here to read the latest release notes.
- 2010-05-28: HapMap3 CNV Genotypes**  
Copy Number Variation genotypes for HapMap phase samples are available for bulk download.
- 2009-12-10: Corrected HapMap3 phased haplotypes available for chromosome X**  
Phased haplotypes for consensus HapMap3 release 2 data for chromosome X has been corrected and the new data are now available for bulk download. Sorry for any inconvenience this might have caused.

9. Click on "HapMap Genome Browser release #28 (Phases 1,2 &3-merged genotypes & frequencies)"



**International HapMap Project**

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**SNP info:** refSNP rs662 with alleles A/G in dbSNP b126 (dbSNP report | Ensembl SNPView)

**Genomic location:** chr7:94775382..94775382, (+) strand relative to the human reference sequence

**Frequency report:**

Population	Genotype frequencies						Allele frequencies										
	genotype	freq	count	genotype	freq	count	Total	Ref-allele	allele	freq	count	Other-allele	allele	freq	count	Total	
ASW (A)	T/T	0.175	10	C/T	0.404	23	C/C	0.421	24	57	T	0.377	43	C	0.623	71	114
CEU (C)	T/T	0.434	49	C/T	0.489	53	C/C	0.097	11	113	T	0.668	151	C	0.332	75	226
CHB (H)	T/T	0.161	22	C/T	0.489	67	C/C	0.350	48	137	T	0.405	111	C	0.595	163	274
CHD (D)	T/T	0.120	13	C/T	0.472	51	C/C	0.407	44	108	T	0.356	77	C	0.644	139	216
GIH (I)	T/T	0.446	45	C/T	0.396	40	C/C	0.158	16	101	T	0.644	130	C	0.356	72	202
JPT (J)	T/T	0.071	8	C/T	0.478	54	C/C	0.451	61	113	T	0.310	70	C	0.690	156	226
LWK (L)	T/T	0.073	8	C/T	0.264	29	C/C	0.664	73	110	T	0.205	45	C	0.795	175	220
MEX (M)	T/T	0.241	14	C/T	0.517	30	C/C	0.241	14	58	T	0.500	58	C	0.500	58	116
MKK (K)	T/T	0.122	19	C/T	0.468	73	C/C	0.410	64	156	T	0.356	111	C	0.644	201	312
TSI (T)	T/T	0.578	69	C/T	0.363	37	C/C	0.059	6	102	T	0.780	155	C	0.220	43	204
YRI (Y)	T/T	0.034	5	C/T	0.374	55	C/C	0.592	87	147	T	0.221	65	C	0.779	229	294

Note: the "reference" allele is the base observed in the reference genome sequence at this location.

**Population descriptors:**  
 ASW (A): African ancestry in Southwest USA  
 CEU (C): Utah residents with Northern and Western European ancestry from the CEPH collection  
 CHB (H): Han Chinese in Beijing, China  
 CHD (D): Chinese in Metropolitan Denver, Colorado  
 GIH (I): Guadalupe Indians in Houston, Texas  
 JPT (J): Japanese in Tokyo, Japan  
 LWK (L): Luhya in Webuye, Kenya  
 MEX (M): Mexican ancestry in Los Angeles, California  
 MKK (K): Maasai in Kinyawa, Kenya  
 TSI (T): Tuscan in Italy  
 YRI (Y): Yoruban in Ibadan, Nigeria

Please see [this page](#) for more information about the populations, as well as a general discussion of the populations under study in the project.

Home | About the Project | Data | Publications | Tutorial  
 Please send questions and comments on website to [hapmap-help@ncbi.nlm.nih.gov](mailto:hapmap-help@ncbi.nlm.nih.gov)

12. Another website to provide more information related to the SNP and the gene  
 UCSC Genome Browser - The UCSC Genome Browser is an interactive website offering access to genome sequence data from a variety of vertebrate and invertebrate species and major model organisms, integrated with a large collection of aligned annotations  
<http://genome.ucsc.edu>

**UCSC Genome Bioinformatics**

Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | Help | About Us

**About the UCSC Genome Bioinformatics Site**

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides portals to ENCODE data at UCSC (2003 to 2012) and to the Neanderthal project. You may download or purchase the Genome Browser source code, or the Genome Browser in a Box (GBiB) at our [online store](#).

We encourage you to explore these sequences with our tools. The [Genome Browser](#) zooms and scrolls over chromosomes, showing the work of annotators worldwide. The [Gene Sorter](#) shows expression, homology and other information on groups of genes that can be related in many ways. [Blat](#) quickly maps your sequence to the genome. The [Table Browser](#) provides convenient access to the underlying database. [VizGene](#) lets you browse through a large collection of in situ mouse and frog images to examine expression patterns. [Genome Graphs](#) allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the [UC Santa Cruz Genomics Institute](#) at the University of California Santa Cruz (UCSC). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#). The Genome Browser is for research use only. Not intended for clinical use.

The Genome Browser project team relies on public funding to support our work. Donations are welcome — we have many more ideas than our funding supports! If you have ideas, drop a comment in our [suggestion box](#).

**News** [Twitter](#) [Facebook](#) [RSS](#) [News Archives](#)

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list. Please see our [blog](#) for posts about Genome Browser tools, features, projects and more.

**10 May 2016 - New Gateway Page!**

The UCSC Genome Browser team is proud to announce a newly redesigned [Genome Browser Gateway](#) page. The Gateway retains its original functionality as a central access point for all genome assemblies available on our site, while sporting several helpful new features and updates:

- Autocomplete searching for any genome browser, genome version, or public hub
- Species browsing through a scrollable visual "tree" menu based on phylogenetic order
- Quick access shortcuts to popular browsers
- New style and color scheme

Please see our [Gateway video](#) for an introduction to these new features.

The new Gateway addresses the need for streamlined access to the rapidly growing number of genome assemblies available on our public site. As an alternative to scrolling through long drop-down menus of genome assemblies, you can now simply type in assembly search terms for autocomplete recognition, visually scroll through our collection, or click a button to access our most popular species.

Note that a few browser utilities that were previously accessed through links and buttons on the Gateway page have been moved to the top menu bar:

- Browser reset: [Genome Browser > Reset All User Settings](#)

13. Click on Genome Browser on top left side

<http://genome.ucsc.edu/cgi-bin/hgGateway>

**UCSC Genome Browser Gateway**

Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

**Browse/Select Species**

POPULAR SPECIES: Human, Mouse, Rat, Fruitfly, Worm, Yeast

REPRESENTED SPECIES: Human, Chimpanzee, Bonobo, Gorilla, Orangutan, Gibbon, Crab-eating macaque, Rhesus, Baboon (anubis), Baboon (hamadryas), Marmoset, Squirrel monkey, Tarsier, Mouse lemur, Bushbaby, Mouse, Rat, Chinese hamster, Kangaroo-rat, Squirrel, Naked mole-rat, Guinea pig, Rabbit, etc.

**Find Position**

Human Assembly  
Feb. 2009 (GRCh37/hg19)

Position/Search Term:  **GO**

Current position: chr7:94,937,196-94,937,696

**Human Genome Browser - hg19 assembly** [view sequences](#)

The February 2009 human reference sequence (GRCh37) was produced by the Genome Reference Consortium. For more information about this assembly, see GRCh37 in the NCBI Assembly database.

**Sample position queries**

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the User's Guide for more information.

Request:	Genome Browser Response:
chr7	Displays all of chromosome 7
chrUn_gI000212	Displays all of the unplaced contig gI000212
20p13	Displays region for band p13 on chr 20
chr3:1-1000000	Displays first million bases of chr 3, counting from p-arm telomere
chr3:1000000-2000	Displays a region of chr3 that spans 2000 bases, starting with position 1000000
RH18061:RH80175	Displays region between genome landmarks, such as the STS markers RH18061 and RH80175, or chromosome bands 15q11 to 15q13, or SNPs rs1042522 and rs1800370. This syntax may also be used for other range queries, such as between uniquely determined ESTs, mRNAs, refSeqs, etc.
15q11:15q13	
rs1042522;rs1800370	
D16S3046	Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well.
AA205474	Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17
AC008101	Displays region of clone with GenBank accession AC008101
AF083811	Displays region of mRNA with GenBank accession number AF083811
PRNP	Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP
NM_017414	Displays the region of genome with RefSeq identifier NM_017414
NP_059110	Displays the region of genome with protein accession number NP_059110

[http://genome.ucsc.edu/cgi-bin/hgTracks?hgsid=496057227\\_PKQc0zA9zCaLbrurq5ywaasjcGEA&org=Human&db=hg19&position=rs662&pix=1052](http://genome.ucsc.edu/cgi-bin/hgTracks?hgsid=496057227_PKQc0zA9zCaLbrurq5ywaasjcGEA&org=Human&db=hg19&position=rs662&pix=1052)

Human rs662 - UCSC Genome Browser v332

genome.ucsc.edu/cgi-bin/hgTracks?hgsid=496057227\_PKQc0zA9zCaLbrurq5ywaasjcGEA&org=Human&db=hg19&position=rs662&pix=1052

**Simple Nucleotide Polymorphisms (dbSNP 146)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 146) Found in >= 1% of Samples**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 144) Found in >= 1% of Samples**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 144)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 142) Flagged by dbSNP as Clinically Assoc**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 142)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 141) Found in >= 1% of Samples**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 141)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**Simple Nucleotide Polymorphisms (dbSNP 138)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

**HapMap SNPs from the CEU Population (Northern and Western European Ancestry in Utah, US - CEPH)**  
[rs662\\_at\\_chr7:94937196-94937696](#)

Click on the first rs662



Enter PON1

<http://www.genecards.org/cgi-bin/carddisp.pl?gene=PON1>

**GeneCards** HUMAN GENE DATABASE

**PON1 Gene (Protein Coding)**  
Paraoxonase 1

Jump to section: Aliases, Disorders, Domains, Drugs, Expression, Function, Genomics, Localization, Orthologs, Paralogs, Pathways, Products, Proteins, Publications, Sources, Summaries, Transcripts, Variants

**Aliases for PON1 Gene**

Paraoxonase 1 <sup>2,3</sup>	Serum Aryldialkylphosphatase <sup>3</sup>
Serum Aryldialkylphosphatase 1 <sup>2,4</sup>	Arylesterase B-Type <sup>3</sup>
Aromatic Esterase 1 <sup>2,4</sup>	Paraoxonase B-Type <sup>3</sup>
Arylesterase 1 <sup>2,3</sup>	EC 3.1.1.81 <sup>4</sup>
A-Esterase 1 <sup>2,4</sup>	EC 3.1.1.2 <sup>4</sup>
Esterase A <sup>2,3</sup>	EC 3.1.8.1 <sup>4</sup>
PON 1 <sup>2,4</sup>	MVCG5 <sup>3</sup>
K-45 <sup>2,4</sup>	ESA <sup>3</sup>
PON <sup>2,4</sup>	

**External IDs for PON1 Gene**  
HGNC: 9204 Entrez Gene: 5444 Ensembl: ENSG0000005421 OMM: 168820 UniProtKB: P27169

**Previous HGNC Symbols for PON1 Gene**  
PON

**Previous GeneCards Identifiers for PON1 Gene**  
GC07M093462, GC07M094525, GC07M094539, GC07M094571, GC07M094764, GC07M094826, GC07M089534

Export aliases for PON1 gene to outside databases

**Summaries for PON1 Gene**

**Entrez Gene Summary for PON1 Gene**

The enzyme encoded by this gene is an arylesterase that mainly hydrolyzes paraxon to produce p-nitrophenol. Paraxon is an organophosphorus anticholinesterase compound that is produced in vivo by oxidation of the insecticide parathion. Polymorphisms in this gene are a risk factor in coronary artery disease. The gene is found in a cluster of three related paraoxonase genes at 7q21.3. (provided by RefSeq, Oct 2008)

**Research Products** for PON1 Gene: Antibodies, Proteins, More...

**Entrez Gene Summary for PON1 Gene**

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**GeneCards Summary for PON1 Gene**

PON1 (Paraoxonase 1) is a Protein Coding gene. Diseases associated with PON1 include microvascular complications of diabetes 5 and aortic valve insufficiency. Among its related pathways are Metabolism and Paroxetone Pathway, Pharmacokinetics. GO annotations related to this gene include calcium ion binding and phospholipid binding. An important paralog of this gene is PON2.

**UniProtKB/Swiss-Prot for PON1 Gene** PON1\_HUMAN,P27169

Hydrolyzes the toxic metabolites of a variety of organophosphorus insecticides. Capable of hydrolyzing a broad spectrum of organophosphate substrates and lactones, and a number of aromatic carboxylic acid esters. Mediates an enzymatic protection of low density lipoproteins against oxidative modification and the consequent series of events leading to atheroma formation.

**Gene Wiki entry for PON1 Gene**

No data available for Tools Summary, PharmGKB "VIP" Summary, IRMAdb sequence ontologies and pRNA Summary for PON1 Gene

**Genomics for PON1 Gene**

**Products:** Regulatory Element

**Regulatory Elements for PON1 Gene**

Transcription factor binding sites by QIAGEN in the PON1 gene promoter: p53 Sp1 MZF-1 Evi-1 AM,1a PPAR-gamma2 PPAR-gamma1 HNF-4alpha1

See All at QIAGEN

**Regulatory Element Products**

SwitchGear PON1 promoter sequence. See all 2 > Browse SwitchGear Promoter luciferase reporter plasmids

**Genomic Location for PON1 Gene**

Chromosome: 7  
Start: 86,297,676 bp from pter End: 86,324,707 bp from pter  
Size: 27,032 bases Orientation: Minus strand

**Genomic View for PON1 Gene**

UCSC Golden Path with GeneCards custom track  
Cytogenetic band: 7q21.3 by Ensembl 7q21.3 by Entrez Gene 7q21.3 by HGNC

GeneDense

**RefSeq DNA sequences for PON1 Gene**

NC\_000007.14 NC\_018918.2 NT\_007933.16

**Proteins for PON1 Gene**

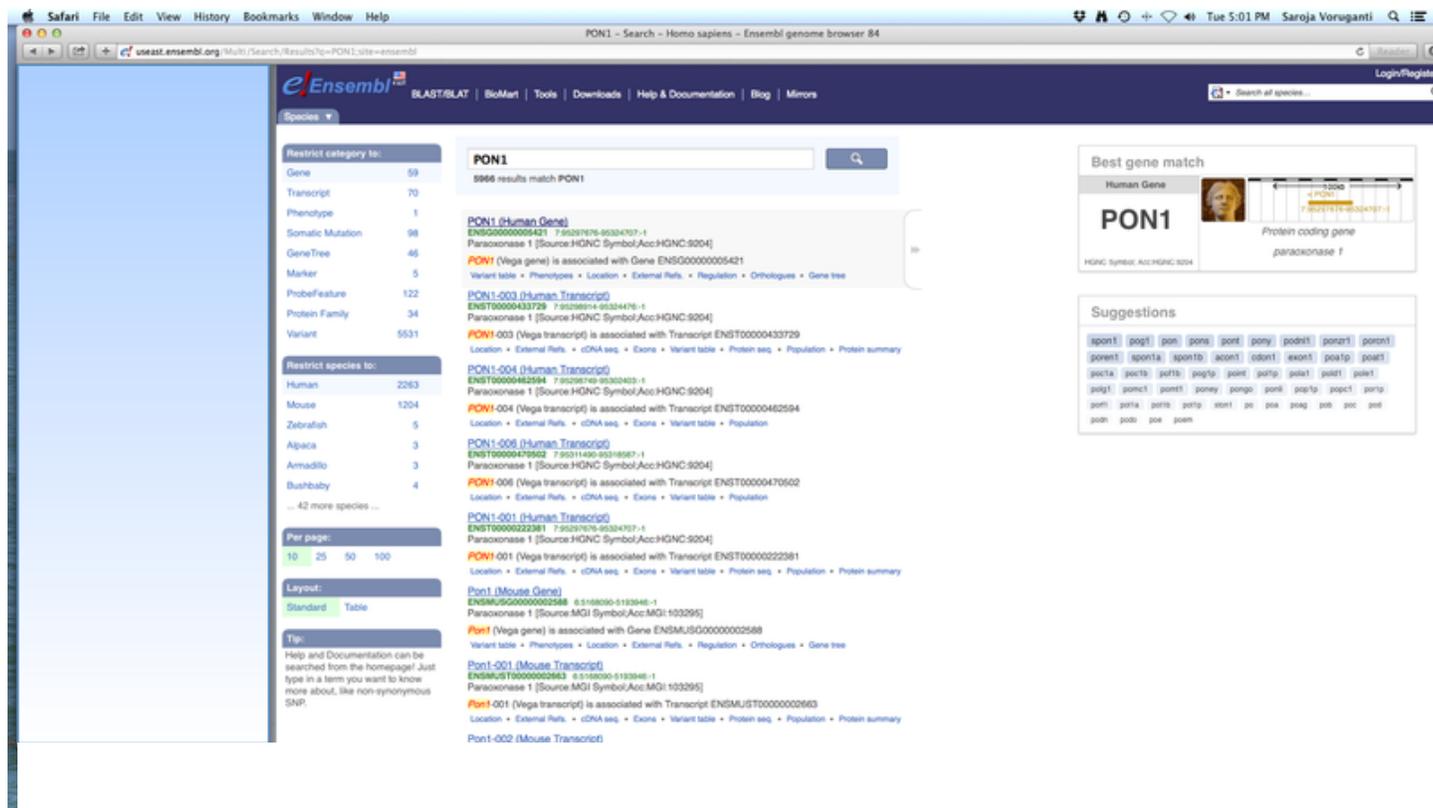
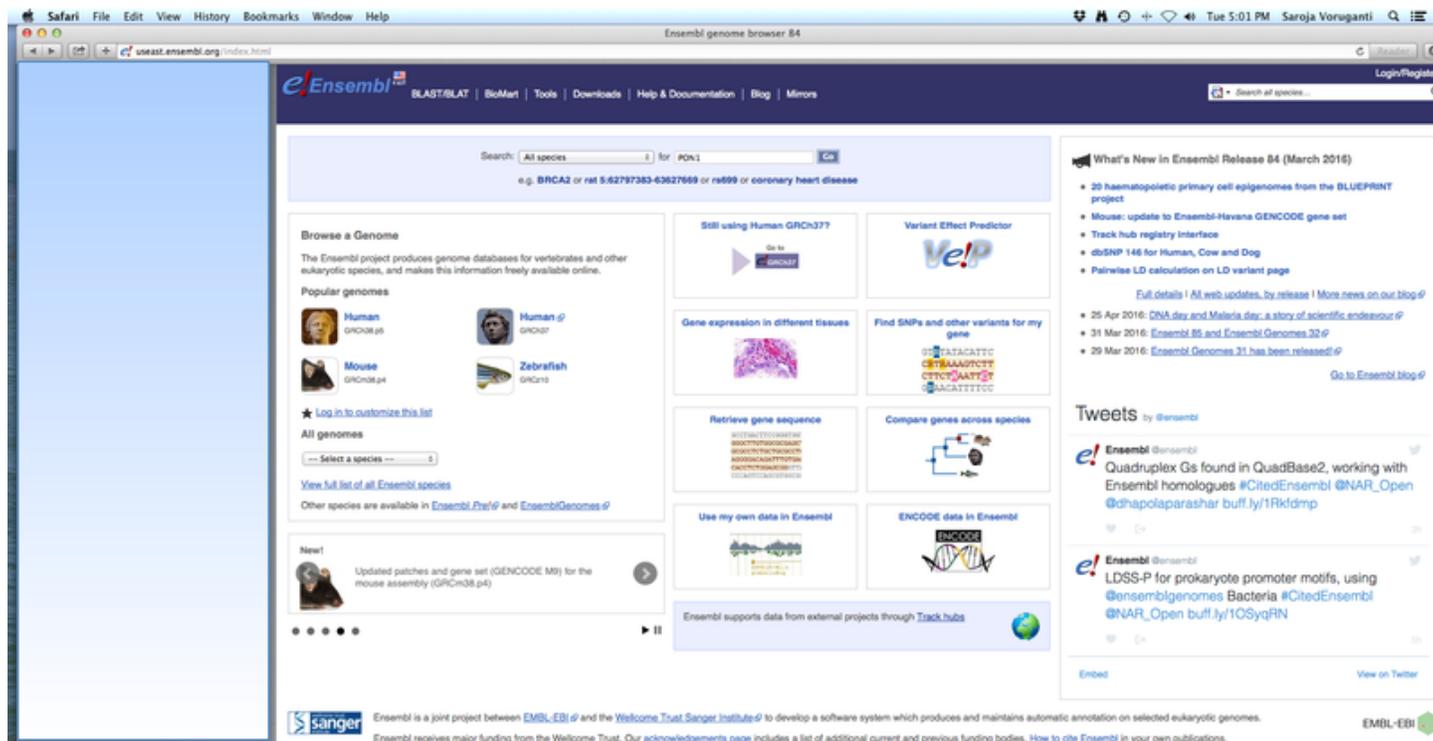
**Products:** Antibody / Protein / Assay

**Protein details for PON1 Gene (UniProtKB/Swiss-Prot)**

One more "Ensembl" <http://useast.ensembl.org/index.html> - Ensembl's aim is to provide a centralized resource for geneticists, molecular biologists and other researchers studying the genomes of our own species and other

vertebrates and model organisms. Ensembl is one of several well known genome browsers for the retrieval of genomic information.

Enter PON1 in the search panel



Click on *PON1* Human gene

**Gene: PON1** (ENSG00000005421)

**Description:** paraoxonase 1 [Source:HGNC Symbol;Acc:HGNC:9204]

**Synonyms:** ESA, PON, MVCD5

**Location:** Chromosome 7: 95,297,676-95,324,702 reverse strand.  
GRCh38:CM000669.2

**About this gene:** This gene has 4 transcripts (splice variants), 73 orthologues, 2 paralogues, is a member of 1 Ensembl protein family and is associated with 3 phenotypes.

**Summary:**

- Name:** [PON1](#) (HGNC Symbol)
- CCDS:** This gene is a member of the Human CCDS set: [CCDS5638.1](#)
- UniProtKB:** This gene has proteins that correspond to the following UniProt identifiers: [P27189](#)
- RefSeq:** Overlapping RefSeq Gene ID [5644](#) matches and has similar biotype of protein\_coding
- Ensembl version:** ENSG00000005421.8
- Other assemblies:** This gene maps to [94,928,988-94,954,019](#) in GRCh37 coordinates. View this locus in the GRCh37 archive: [ENSG00000005421.8](#)
- Gene type:** Known protein coding
- Annotation method:** Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).
- Alternative genes:** This gene corresponds to the following database identifiers: Havana gene: [DTX:HUNG0000153895](#)

**Gene-based displays:** Summary, Splice variants, Transcript comparison, Supporting evidence, Gene alleles, Sequence, Secondary Structure, External references, Regulation, Ontologies, Comparative Genomics, Gene tree, Gene gain/loss tree, Orthologues, Paralogues, Ensembl protein families, Phenotype, Genetic Variation, Variant table, Variant image, Structural variants, External data, ID History, Gene history.

**Go to Region in Detail for more tracks and navigation options (e.g. zooming)**

**Genes (Comprehensive):** PIP DSA-005, PIP DSA-202, PIP DSA-001, PIP DSA-002, PIP DSA-003

**Gene type:** Known protein coding

**Annotation method:** Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).

**Alternative genes:** This gene corresponds to the following database identifiers: Havana gene: [DTX:HUNG0000153895](#)

**Go to Region in Detail for more tracks and navigation options (e.g. zooming)**

**Genes (Comprehensive):** PIP DSA-005, PIP DSA-202, PIP DSA-001, PIP DSA-002, PIP DSA-003

**Transcripts (Comprehensive):** PIP DSA-005, PIP DSA-202, PIP DSA-001, PIP DSA-002, PIP DSA-003

**Gene Legend:** Protein Coding (red), Ensembl protein coding (orange), merged Ensembl/Havana (yellow), Non-Protein Coding (blue), processed transcript (green)

**Configuring the display:** Tip: use the "Configure this page" link on the left to show additional data in this region.

Ensembl release 84 - March 2016 © WT3 / EMBL-EBI [Permanent link - View in archive site](#)

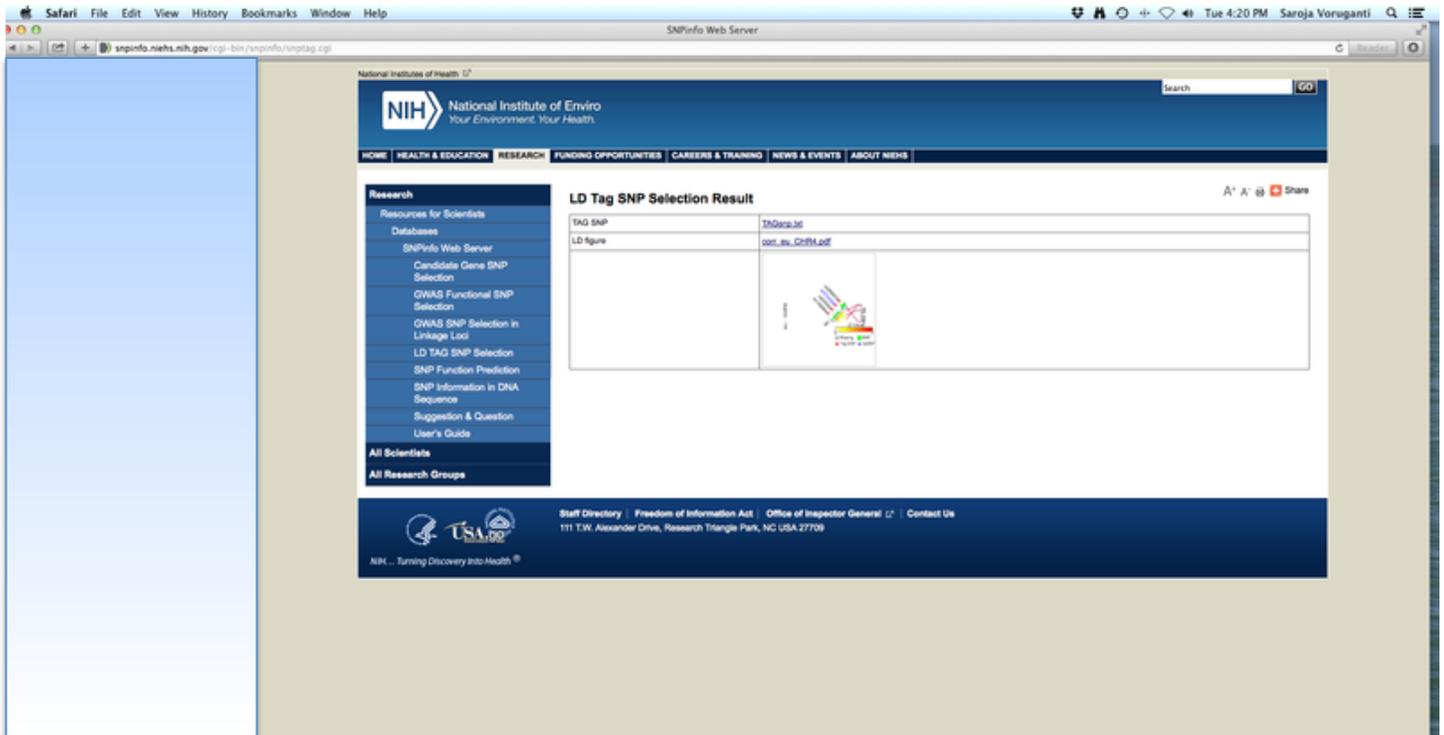
For information on LD and Tag SNPs <https://snpinfo.niehs.nih.gov/snpinfo/snptag.htm> - SNPinfo is designed to comprehensively utilize computational (predicted functional SNPs that have differential affect between reference allele and alternative allele), experimental and epidemiological information together with recent genome wide association study

(GWAS) results and linkage disequilibrium (LD) information to prioritize SNPs for further genetic mapping studies

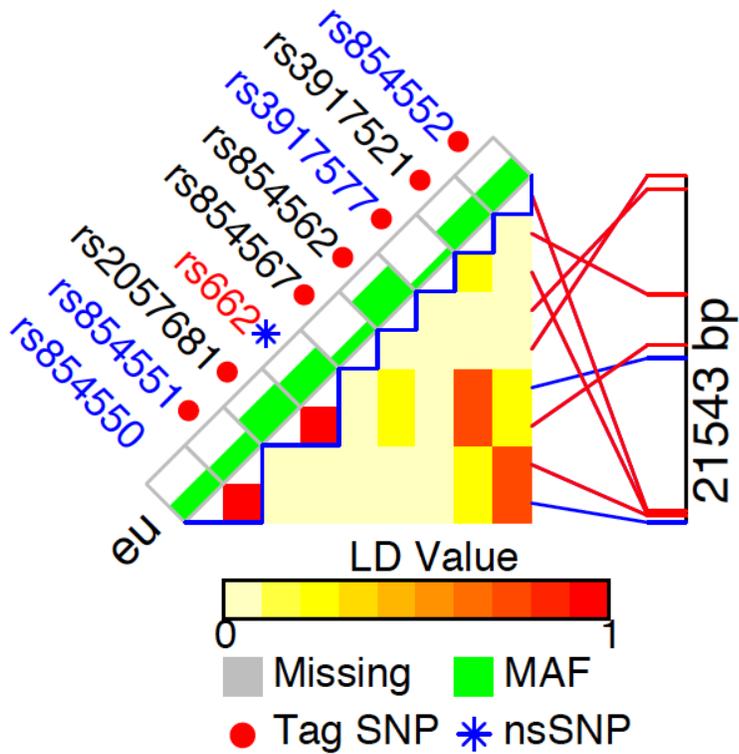
Select SNPrsID if you are entering SNP ID. I selected SNP rsID  
Select either dbSNP or HapMap. I selected dbSNP

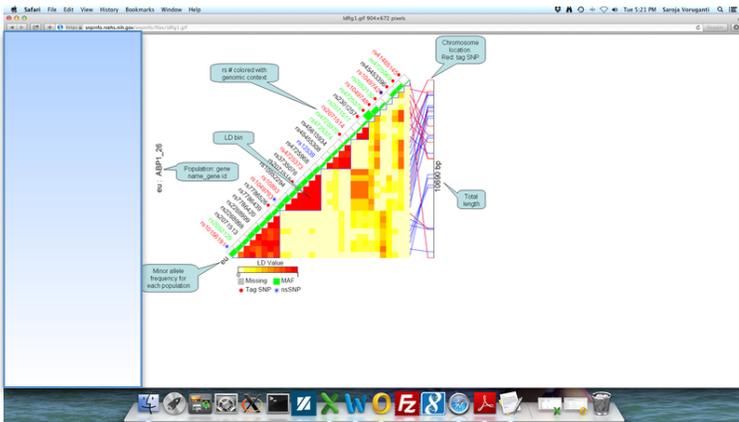
The screenshot shows the NIH website header with the logo and navigation menu. On the left is a sidebar with a 'Research' menu where 'LD TAG SNP Selection' is highlighted. The main content area is titled 'LD TAG SNP Selection (TagSNP)'. It features a 'Query by:' dropdown set to 'Gene Name'. Below are input fields for 'Gene name', '5' Flanking (bp): 0, and '3' Flanking (bp): 0. The 'Genotype Data:' dropdown is set to 'HapMap'. A 'Population:' section contains checkboxes for various groups: ASW, CEU, CHB, CHD, GIH, JPT, LWK, MEX, MKK, TSI, and YRI. There are also checkboxes for 'Force in SNPs', 'Force out SNPs', 'SNP design score', and 'SNP design score cutoff' (set to 0.6). A second section shows 'LD method' with radio buttons for 'r2' and 'CLD'. Below this are input fields for 'Minimum # of valid genotype pairs required to calculate LD' (set to 5), 'LD threshold' (set to 0), 'Maximum distance (bp) between SNPs for calculation of LD' (set to 250000), 'Minor allele frequency range' (set to 0.05,0.5), and 'Minimum number of SNPs tagged by each tag SNP' (set to 1).

This screenshot shows the same NIH website but with a different configuration in the 'LD TAG SNP Selection (TagSNP)' tool. The 'Query by:' dropdown is now set to 'SNP rsID'. A text area contains a list of SNP IDs: rs2282679, rs791130, rs145971733, and rs14547263355. The 'Genotype Data:' dropdown is set to 'dbSNP'. The 'Population:' section has checkboxes for 'Sub-Saharan African', 'Asian', 'European', 'African American', and 'Hispanic', with 'European' selected. The 'SNP design score cutoff' is now set to 0.6. The 'LD method' section remains the same with 'r2' selected. The other parameters (minimum genotype pairs, LD threshold, maximum distance, minor allele frequency range, and minimum number of SNPs tagged) are also the same as in the first screenshot.



Click on the pdf link and you will get the LD plot for *PON1* SNPs



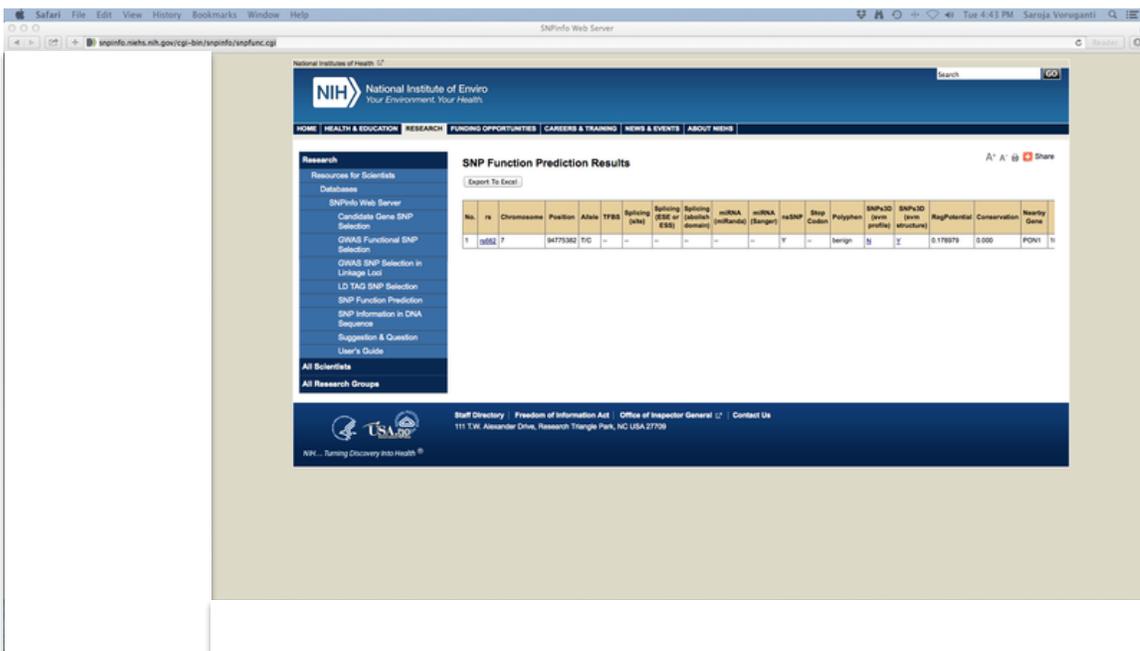


10. For functional significance of rs662 click on SNP function prediction

<https://snpinfo.niehs.nih.gov/snpinfo/snfunc.htm>

enter rs662 in “paste a list of SNP rsid panel” and select a population

<https://snpinfo.niehs.nih.gov/cgi-bin/snpinfo/snfunc.cgi>



### FuncPred: Functional SNP Prediction

SNPs may have functional effects including: transcriptional regulation by affecting transcription factor binding sites (TFBS) activity; premature termination of amino-acid sequence (stop codons); changing of splicing pattern or efficiency by disrupting splice site, exonic splicing enhancers (ESE) or silencers (ESS); alteration of protein structures or properties by changing single amino acids or changing the frame of the protein-coding region; regulation of protein translation by affecting microRNA (miRNA) binding sites activity. Many software tools or web servers can be used to predict TFBSs, ESE site, ESS site or miRNA binding sites. However, although SNPs may be located in such binding sites, the alternative alleles of a SNP may not necessarily have different activities. We designed several pipelines to predict SNPs that may affect biological function with alternative alleles.

### Include SNPs with LD > ... in population

Display SNPs that are in high LD (in population selected) with SNPs in the query SNP list.

## nsSNP

SNPs in protein-coding regions that can cause amino acid change (non-synonymous coding SNPs, nsSNP).

## Stop Codon

SNPs that may lead to premature termination of peptides (non-sense), which would disable the protein function.

**Polyphen prediction** [Polyphen](http://genetics.bwh.harvard.edu/pph/) (<http://genetics.bwh.harvard.edu/pph/>) (Sunyaev, Ramensky et al. 2001) method predicted damaging nsSNPs.

**SNPs3D prediction** [SNPs3D](http://www.snps3d.org/) (<http://www.snps3d.org/>) (Yue, Melamud et al. 2006) method predicted damaging nsSNPs.

## TFBS Prediction

If a non-coding SNP is located at a transcription factor-binding site (TFBS) of a gene, then it may affect the level, location, or timing of gene expression. We predicted such SNPs according to the procedure described in Xu and Taylor (submitted).

**Refine TFBS** Several studies (Elnitski et al 2003,2006; King et al, 2005) show that using both the predicted conserved TFBS together with the regulatory potential score (RP Score) (downloaded from [UCSC genome bioinformatics web site](http://genome.ucsc.edu) (<http://genome.ucsc.edu>)) can result in more precise predictions, so we also provide this option on the web server.

## Splicing regulation

SNPs that are located at 2 base pair of intron-exon junction region, exonic splicing enhancer (ESE), or exonic splicing silencer (ESS) may disrupt splicing activity and cause alternative splicing. We predict SNPs whose alternative alleles may affect splicing using the methods detailed in Xu and Taylor (submitted).

## miRNA Binding Site Prediction

microRNAs (miRNA) are single-stranded RNA molecules of about 21-23 nucleotides in length, which can inhibit protein translation through binding to the end of a messenger RNA (mRNAs). We predicted SNPs that may affect miRNA binding site activity according to the methods described in Xu and Taylor (submitted).

## Regulatory Potential Score

Regulatory potential score (ESPERR Regulatory Potential (7 Species)) downloaded from UCSC genome bioinformatics web site (<http://genome.ucsc.edu/>). Because SNPs in coding region are tend to have higher regulatory potential scores (see Figure, lines for all SNPs in human genome, non-coding SNPs and intron SNPs are overlapped), So we use this score only for SNPs that are outside of coding region in SNP selection.

## Conservation Score

Vertebrate Multiz Alignment and Conservation score (17 Species) downloaded from UCSC genome bioinformatics web site (<http://genome.ucsc.edu/>). Because SNPs in coding region are tend to have higher conservation score (see Figure, lines for all SNPs in human genome, non-coding SNPs and intron snps are overlapped), so we use this score only for SNPs that are outside of the coding region of genes in SNP selection.

For more LD information

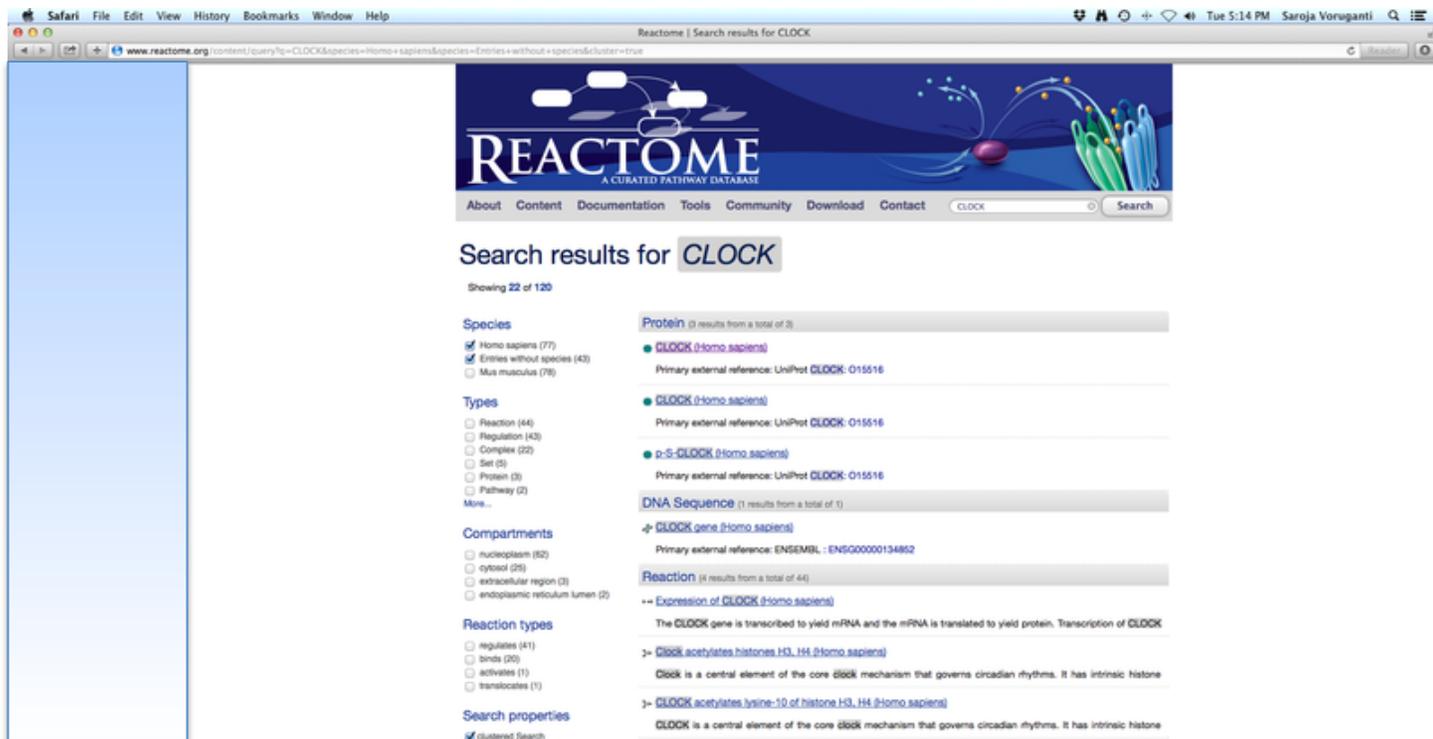
<https://www.broadinstitute.org/mpg/snap/> - SNAP finds proxy SNPs based on linkage disequilibrium, physical distance and/or membership in selected commercial genotyping arrays. Pair-wise linkage disequilibrium is pre-calculated based on phased genotype data from the [International HapMap Project](#). Information about the genotyping arrays is based on data published by the vendors. SNAP can also generate linkage disequilibrium plots, like the one shown at the right. To generate plots, click on the Plots tab above and select plotting options.



Reactome  
<http://www.reactome.org> - a free online curated pathway database



enter CLOCK in the search panel



Click on CLOCK (Homo Sapiens)

Safari File Edit View History Bookmarks Window Help Reactome | CLOCK

www.reactome.org/content/detail/R-HSA-879828

# REACTOME

A CURATED PATHWAY DATABASE

About Content Documentation Tools Community Download Contact e.g. DR5431, NTN1, signaling by Search

## ● CLOCK (R-HSA-879828) [Homo sapiens]

Reference Gene Product

Locations in the PathwayBrowser

- ↕ Circadian Clock (Homo sapiens)
- ↕ Chromatin organization (Homo sapiens)

Other forms of this molecule

CLOCK (lysine)      p-S-CLOCK (nucleoplasm)

Additional Information

External reference name	CLOCK																																			
External reference id	Q15516																																			
Synonyms	Circadian locomotor output cycles protein kaput																																			
Compartment	nucleoplasm																																			
Other Identifiers	<table border="1"> <tr> <td>000390253</td> <td>11727626_x_at</td> <td>11749633_x_at</td> <td>204980_at</td> <td>229856_at</td> </tr> <tr> <td>227521_at</td> <td>30080_at</td> <td>4915</td> <td>51997_at</td> <td>51998_at</td> </tr> <tr> <td>784_x_at</td> <td>9175</td> <td>A_23_P419038</td> <td>A_24_P158186</td> <td>A_33_P3358938</td> </tr> <tr> <td>AA83969</td> <td>AA113733</td> <td>AA26158</td> <td>AA26160</td> <td>AB002332</td> </tr> <tr> <td>AB002332_at</td> <td>AB005535</td> <td>AB005535_x_at</td> <td>ABM4208</td> <td>AF011568</td> </tr> <tr> <td>AI008442</td> <td>AK091708</td> <td>BA020782</td> <td>BA021774</td> <td>BAF64397</td> </tr> <tr> <td>BC126137</td> <td>BC126139</td> <td>CC053500</td> <td>CLOCK</td> <td>CLOCK-001</td> </tr> </table>	000390253	11727626_x_at	11749633_x_at	204980_at	229856_at	227521_at	30080_at	4915	51997_at	51998_at	784_x_at	9175	A_23_P419038	A_24_P158186	A_33_P3358938	AA83969	AA113733	AA26158	AA26160	AB002332	AB002332_at	AB005535	AB005535_x_at	ABM4208	AF011568	AI008442	AK091708	BA020782	BA021774	BAF64397	BC126137	BC126139	CC053500	CLOCK	CLOCK-001
000390253	11727626_x_at	11749633_x_at	204980_at	229856_at																																
227521_at	30080_at	4915	51997_at	51998_at																																
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AA83969	AA113733	AA26158	AA26160	AB002332																																
AB002332_at	AB005535	AB005535_x_at	ABM4208	AF011568																																
AI008442	AK091708	BA020782	BA021774	BAF64397																																
BC126137	BC126139	CC053500	CLOCK	CLOCK-001																																
Secondary Identifiers	CLOCK_HUMAN, ADAV01, AZ019, O14516, GBUT8																																			
Gene Names	CLOCK, BHLHE8, KAA0334																																			
Chain	chain:1-848																																			

This entry is a component of:

Click on Circadian Clock (Homo Sapiens)

Safari File Edit View History Bookmarks Window Help Reactome | PB | Circadian Clock

www.reactome.org/PathwayBrowser/#/R-HSA-400253

REACTOME 10.3.2

Pathways for: Homo sapiens

Analysis Tour Layout

Cell Cycle

- Cell-Cell communication
- Cellular responses to stress
- Chromatin organization
- Circadian Clock**
- Developmental Biology
- Disease
- DNA Repair
- DNA Replication
- Extracellular matrix organization
- Gene Expression
- Homeostasis
- Immune System
- Autophagy
- Metabolism
- Metabolism of proteins
- Muscle contraction
- Neuronal System
- Organelle biogenesis and maintenance
- Programmed Cell Death
- Reproduction
- Signal Transduction
- Transmembrane transport of small molecules
- Vesicle-mediated transport

Description

Circadian Clock    Species: Homo sapiens

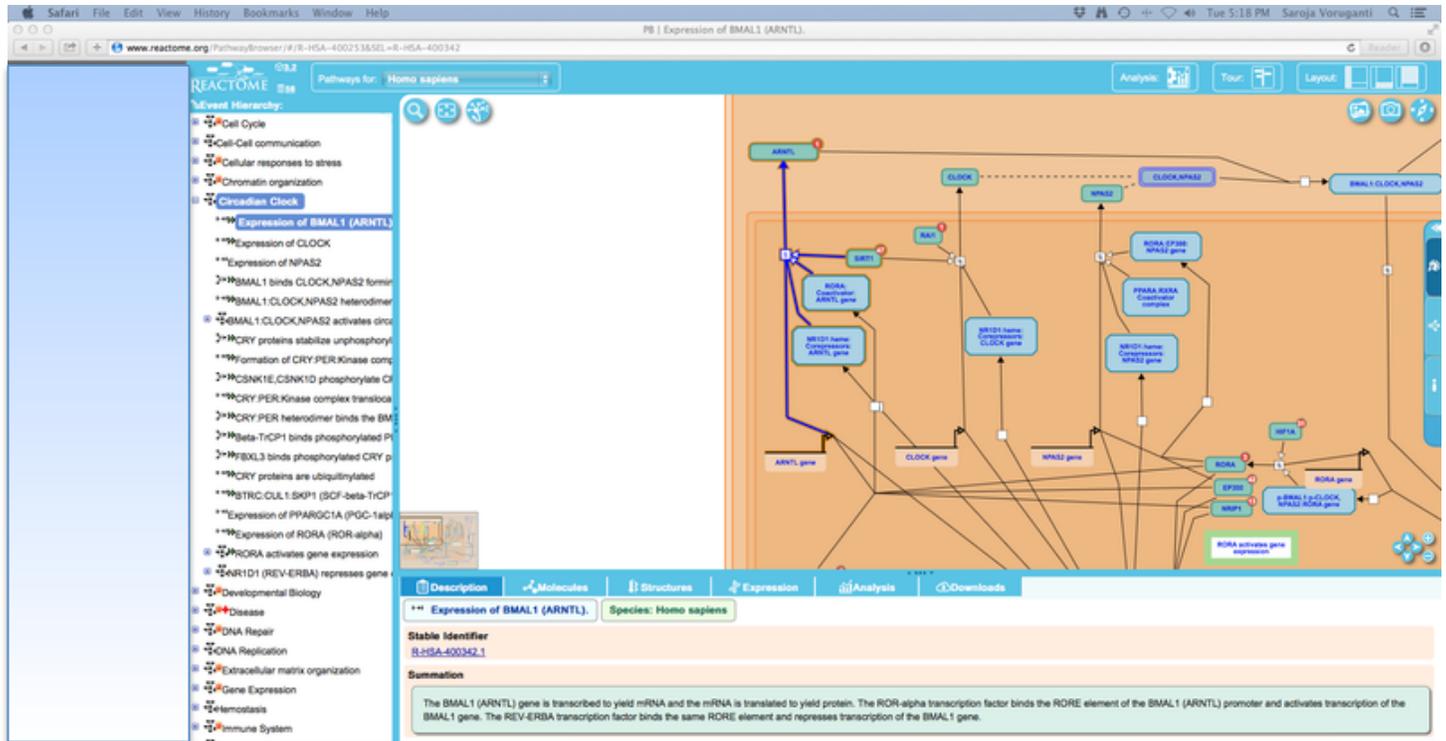
DOI: 10.31801/REACT\_24941.1

Stable Identifier: R-HSA-400253.1

Summary

At the center of the mammalian circadian clock is a pacemaker transcription-translation-based feedback loop: The BM1/1 (CLOCK/BMAL1) (BM1/1 (CLOCK/BMAL1) heterodimer transactivates PER2 and PER3 genes to produce E. An

you can click on Circadian Clock on the left side and click on each subsection to uncover the pathways. First one is shown as an example



Mauche S and Schunkert H. Strategies beyond genome-wide association studies for atherosclerosis. *Arterioscler Thromb Vasc Biol.*2012;32:170-181

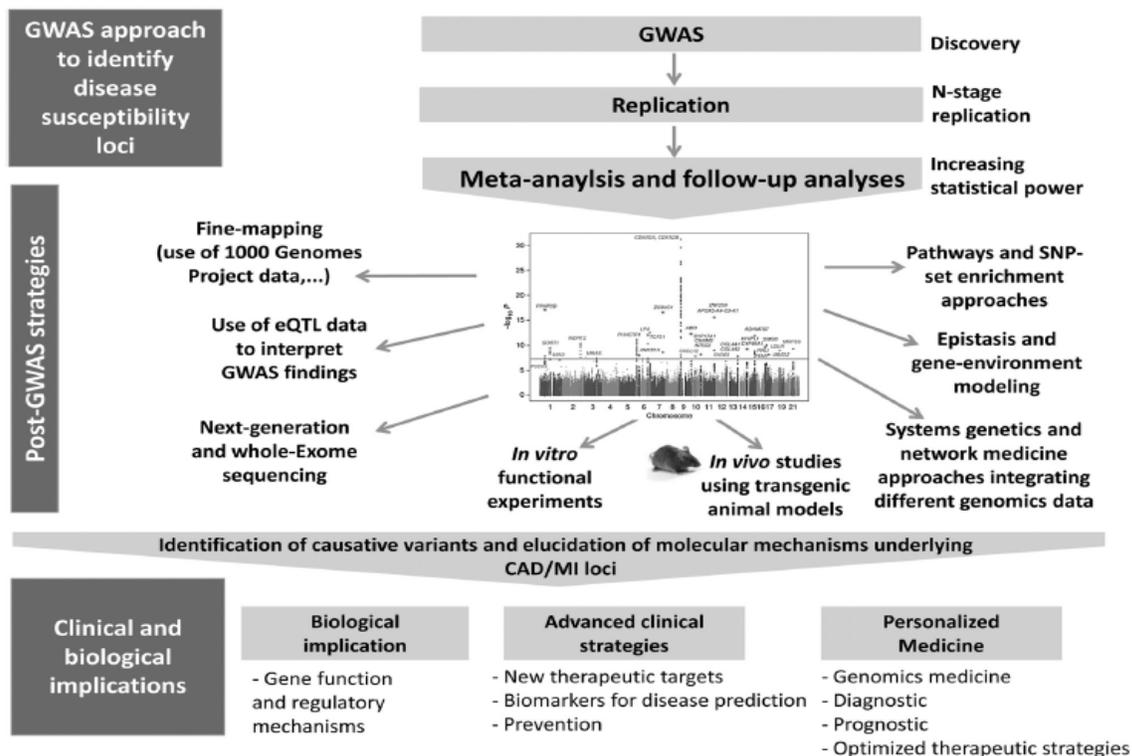


Figure: Post-genome-wide association study strategies and biological and clinical implications of GWAS findings.