

metabolicMine.org

Mike Lyne
Adrian Carr
Richard Smith
Radek Stepan
Gos Micklem



Systems Biology Centre,
University of Cambridge

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metabolicMine.org

45 minutes + 15...

- Team of ~7 since 2002
- FlyMine - www.flymine.org
 - 30+ data sources, *Drosophila* & *Anopheles*
- Yeast, Rat, Zebrafish MODs (interoperation)
 - ratmine.mcw.edu
- modENCODE - intermine.modencode.org
 - whole genome scale *C. elegans*/*D. melanogaster*

InterMine is generic software used to build the above



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Developed for Fly, used for yeast, rat, zebrafish + modENCODE + maybe worm/mouse.
InterMine is generic core.

A lot of the features developed for the above are appropriate for medical focus.

With a view to making metabolic disease version did survey...

metabolicMine Objectives

Reach information without having to visit several sites and formats

- Examine properties of a collection
- Investigate gene function
- Evaluate individual sequence variations

- Genome region searches
- Work with old /diverse identifiers
- Export in a range of formats
- Link to other tools



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Different sites & formats: extremely time consuming!
collection of genes/SNPs
InterMine is well suited to address these needs

Data Sources

- Search across information from many different sources and types including, Genes, Proteins, Diseases, SNPs, GWAS, Homologues, Pathways, Interactions and more!




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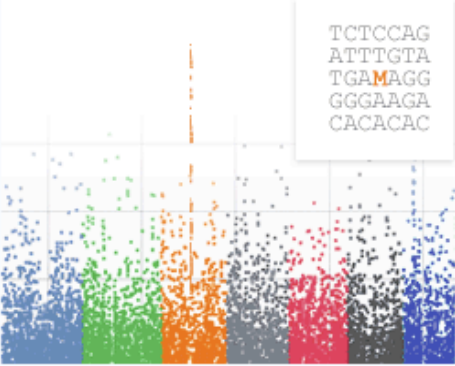
Experience with integration of data from varied sources & formats


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Data mining metabolic diseases

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First time here?

Welcome to **metabolicMine**, an integrated web resource of Data & Tools to support the Metabolic Disease research community.

If you are short of time, just navigate through our set of [Feature Hints](#). For a basic overview of the site and its features try the [Quick Tour](#), it takes about ten minutes.

[TAKE A TOUR](#)


Search

Enter a gene, protein, SNP or other identifier [eg. [PPARG](#), [Insulin](#), [rs876498](#)].

Alternatively, search for disease, keywords or publications [eg. [Diabetes](#), [GWAS](#), [PMID](#), [Author](#)]

[SEARCH](#)

Analyse Lists of Data

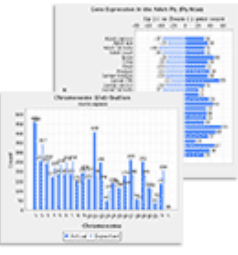


PPARG, ENSG00000108753, FT0, 3949, LEP, 946, MC3R, ENSG00000164326, LPL, LDLR, P55916, ENSG00000118137, GLUT4, ENST00000299766, SLC27A1

[file upload](#)
[ANALYSE LIST](#)

Explore and Analyse.

Upload your own data or browse our Public sets. Covering Pathways to Publications, discover hidden relationships with our analysis widgets.



Use Template Searches

Get started with **powerful searches** using our predefined Templates. These customizable templates have been designed around common tasks performed by our biologist community.

[Genes](#)
[Proteins](#)
[SNPs](#)
[Diseases](#)
[Pathways](#)
[Interactions](#)
[Homology](#)
[GO](#)

metabolicMine loads Gene models and sequence from ENSEMBL. We additional cross-reference Gene information from many additional sources. [Read more](#)

Search for genes:

Region ➡ SNP
Gene ➡ Chromosomal location.
Chromosomal Location ➡ All Genes + Transcripts + Exons

Region ➡ Genes
Gene ➡ SNPs
Gene ➡ Overlapping genes.

» [More queries](#)

popular templates

Integrates broad-scale public data
 Integrates more-specific data sets
 gene expression / proteomics, user / community nominations, own lab data...
 Library of relevant Template Searches
 List Analysis tools / widgets
 public List nominations
 Web services
 query APIs for bioinformaticians
 interoperation with existing tools / databases
 Comparative studies via interoperation with Model Organism Databases

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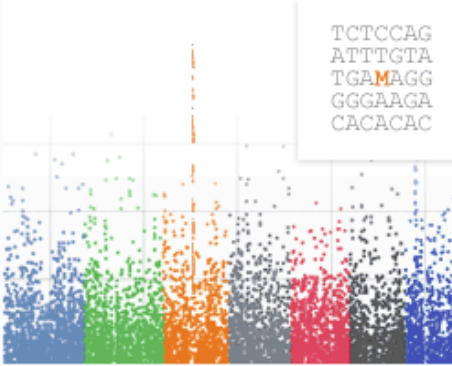
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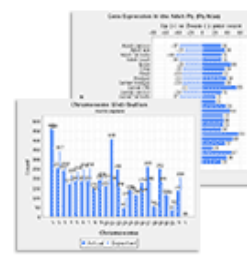


PPARG, ENSG00000108753, FT0, 3949, LEP, 946, MC3R, ENSG00000164326, LPL, LDLR, P55916, ENSG00000118137, GLUT4, ENST00000299766, SLC27A1

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Use Template Searches

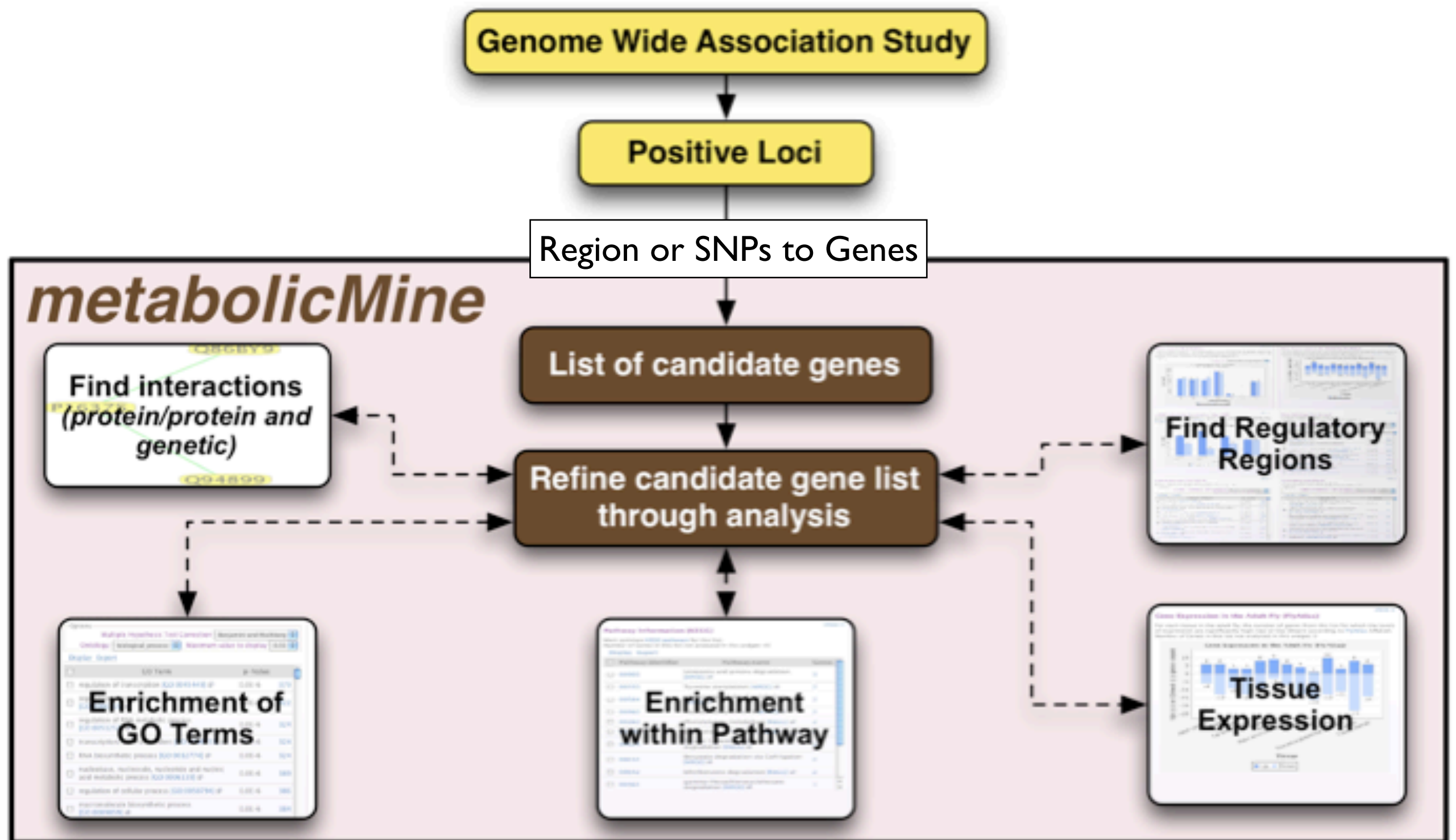
Get started with **powerful searches** using our predefined Templates. These customizable templates have been designed around common tasks performed by our biologist community.

Genes	Proteins	SNPs	Diseases	Pathways	Interactions	Homology	GO
metabolicMine loads Gene models and sequence from ENSEMBL. We additional cross-reference Gene information from many additional sources. Read more							
Search for genes:							
Region → SNP		Region → Genes					
Gene → Chromosomal location.		Gene → SNPs					
Chromosomal Location → All Genes + Transcripts + Exons		Gene → Overlapping genes.					
» More queries							

popular templates

Integrates broad-scale public data
Integrates more-specific data sets
gene expression / proteomics, user / community nominations, own lab data...
Library of relevant Template Searches
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Examine the Properties of a Collection: List Analysis




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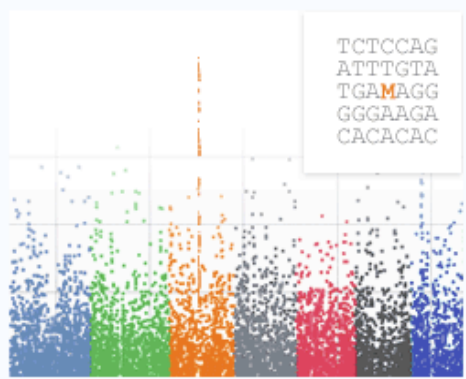
List Analysis provides an overview of your collection across a range of properties including GO terms, Pathways, Interactions, Publications and Expression. Display 'widgets' highlight statistically enriched results.


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[SEARCH](#)



Analyse Lists of Data

Gene

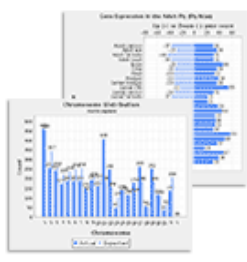
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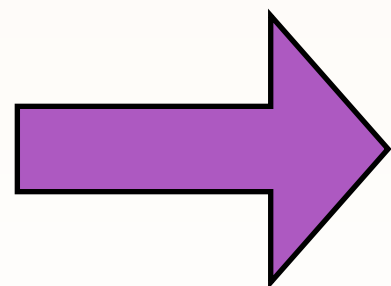
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A Library of 'Template' Searches

Covers questions frequently asked by the research community...

Genes --> Proteins

Genes --> all SNPs

Genes --> Diseases

Gene --> Pathways

Proteins --> Protein Domains

Proteins --> Interactions

Proteins --> GO terms

Protein Domain --> all Proteins

Go term --> Proteins

SNPs --> GWA study

SNPs --> Type + Consequences

GWAS term --> SNPs

GWA study --> Genes

Region --> Genes

Region --> SNPs

Gene A --> Interactions <-- Gene B



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One of key features – ability to search across domains of knowledge

A Library of 'Template' Searches

Covers questions frequently asked by the research community...

Gene A + Gene B → Pathways ☆

For a given Gene A (or List A of Genes) show any associated Pathway(s) in common with Gene B (or List B of Genes). Data Source: KEGG or REACTOME. Keywords: pathways, metabolism, cascade, reaction

Gene A

LOOKUP: for Organism:

☒ constrain to be saved Gene list

Gene B

LOOKUP: for Organism:

☒ constrain to be saved Gene list

[< embed results />](#) [web service URL](#) [Perl](#) | [Python](#) | [Java \[help\]](#) [export XML](#)

'Template' - an interactive Search Form

Candidate Gene List

List of 'known' Disease Genes



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Complex searches are represented as pre-filled Search Forms

**Make a new List
from your results**

**Export Results in
a number of
different formats**

*Access summary data
for each column*

**Click header
to select all
entries ...**

Links to Report pages

... or select individual entries

Total rows: 100
Total unique values: 15

Value	Count
Metabolism of lipids and lipoproteins	36
PPAR signaling pathway	13
GPCR downstream signaling	10
Signaling by GPCR	10
Glycerolipid metabolism	6
Lipid digestion, mobilization, and transport	6
Pathways in cancer	4
Chylomicron-mediated lipid transport	2
Lipoprotein metabolism	2
Neuroactive ligand-receptor interaction	2

For a given Gene A (in List A of Genes) show any associated Pathway, reaction

CREATE LIST ADD TO LIST ADD COLUMN **EXPORT** PAGE SIZE 25

Selected:LPIN1, PLIN1, LIPE, DGKA, CPT1A, CPT2

Gene > symbol	Gene > name	Gene > pathways > name	Gene > pathways > identifier	... pathways > dataSets > name	... pathways > dataSets > symbol	Pathways in cancer
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> GPAM	Chylomicron-mediated lipid transport
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> LDLR	Lipoprotein metabolism
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833		<input checked="" type="checkbox"/> LIPE	Neuroactive ligand-receptor interaction
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833		<input checked="" type="checkbox"/> LPIN1	lipin 1
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833		<input checked="" type="checkbox"/> PLIN1	perilipin 1
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833		<input type="checkbox"/> PLIN2	perilipin 2
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> Metabolism of lipids and lipoproteins	556833		<input type="checkbox"/> PRKAA2	protein kinase, AMP-activated
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> PPAR signaling pathway	03320	<input type="checkbox"/> KEGG pathways data set	<input type="checkbox"/> ADIPOQ	adiponectin, C1Q and collagen
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> PPAR signaling pathway	03320	<input type="checkbox"/> KEGG pathways data set	<input checked="" type="checkbox"/> CPT1A	carnitine palmitoyltransferase
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> PPAR signaling pathway	03320	<input type="checkbox"/> KEGG pathways data set	<input checked="" type="checkbox"/> CPT2	carnitine palmitoyltransferase
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> PPAR signaling pathway	03320	<input type="checkbox"/> KEGG pathways data set	<input checked="" type="checkbox"/> PLIN1	perilipin 1
<input type="checkbox"/> LPL	lipoprotein lipase	<input type="checkbox"/> PPAR signaling pathway	03320	<input type="checkbox"/> KEGG pathways data set	<input type="checkbox"/> SCD	stearoyl-CoA desaturase (delta 5)
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input checked="" type="checkbox"/> DGKA	diacylglycerol kinase, alpha 8C
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> DGKD	diacylglycerol kinase, delta 13
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> DGKQ	diacylglycerol kinase, theta 11
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> PIK3CA	phosphoinositide-3-kinase, class I
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> PIK3CG	phosphoinositide-3-kinase, class I
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> Neuroactive ligand-receptor interaction	04080	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> LEP	leptin
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> Signaling by GPCR	372790	<input type="checkbox"/> Reactome data set	<input checked="" type="checkbox"/> DGKA	diacylglycerol kinase, alpha 8C
<input type="checkbox"/> MC3R	melanocortin 3 receptor	<input type="checkbox"/> Signaling by GPCR	372790	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> DGKD	diacylglycerol kinase, delta 13
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<input type="checkbox"/> MC4R	melanocortin 4 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input checked="" type="checkbox"/> DGKA	diacylglycerol kinase, alpha 8C
<input type="checkbox"/> MC4R	melanocortin 4 receptor	<input type="checkbox"/> GPCR downstream signaling	388396	<input type="checkbox"/> Reactome data set	<input type="checkbox"/> DGKD	diacylglycerol kinase, delta 13

⏪ ⏩ | Displaying rows 51 to 75 | Total rows: 100



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Results are linked to database entries and can be a starting point for further analysis. Create Lists, Export in a number of formats and view summary statistics.

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[Contact Us](#)
Gene: PPARG *H. sapiens*
primaryIdentifier **ENSG00000132170** secondaryIdentifier **5468**
symbol **PPARG** name **peroxisome proliferator-activated receptor gamma**
organism.shortName **H. sapiens** mapLocation **3p25**
ncbiGeneNumber **5468**
synonyms: NR1C3, PPARG1, CIMT1, PPARGgamma, PPARG2, GLM1
[SHARE](#)

Quick Links:

Summary | Genomics | Proteins | SNPs | Disease | Comparative Genomics | Interactions | Literature | Gene Ontology | Other

Genome feature

Sequence ontology type:	gene	Length:	146989	FASTA...
Location:	3:12328867-12475855 forward strand			

Curated comments from UniProt

Type	Comment	Proteins
disease	Defects in PPARG are the cause of familial partial lipodystrophy type 3 (FPLD3) [MIM:604367]. Familial partial lipodystrophies (FPLD) are a heterogeneous group of genetic disorders characterized by marked loss of subcutaneous (sc) fat from the extremities. Affected individuals show an increased preponderance of insulin resistance, diabetes mellitus and dyslipidemia.	PPARG_HUMAN
disease	Defects in PPARG may be associated with susceptibility to obesity [MIM:601665].	PPARG_HUMAN
disease	Genetic variations in PPARG can be associated with susceptibility to glioma type 1 (GLM1) [MIM:137800]. Gliomas are central nervous system neoplasms derived from glial cells and comprise astrocytomas, glioblastoma multiforme, oligodendrogliomas, and ependymomas. Note=Polymorphic PPARG alleles have been found to be significantly over-represented among a cohort of American patients with sporadic glioblastoma multiforme suggesting a possible contribution to disease susceptibility.	PPARG_HUMAN
disease	Note=Defects in PPARG can lead to type 2 insulin resistant diabetes and hypertension. PPARG mutations may be associated with colon cancer.	PPARG_HUMAN
function	Receptor that binds peroxisome proliferators such as hypolipidemic drugs and fatty acids. Once activated by a ligand, the receptor binds to a promoter element in the gene for acyl-CoA oxidase and activates its transcription. It therefore controls the peroxisomal beta-oxidation pathway of fatty acids. Key regulator of adipocyte differentiation and glucose homeostasis.	PPARG_HUMAN
similarity		D2KUA6_HUMAN
similarity		PPARG_HUMAN

Lists

This Gene is in 5 lists:

- PL_BHF_UCL_cardiovascGenes
- PL_monogenGlucTolerance_ORa
- PL_T2Dloci_DorHodgkinLect_Mc
- PL_T2DassocGenes_PharmGKB
- PL_T2DcandGenes_Voight2010

Link to other InterMines

[RatMine](#)
R. norvegicus
[Pparg](#)

External Links

ArrayExpress Atlas: [ENSG00000132170](#)
HuGe: [5468](#)
ensembl: [ENSG00000132170](#)
BioGPS: [5468](#)
HapMap: [ENSG00000132170](#)
Entrez Gene: [5468](#)

Lists which contain this Gene

Links to contextual information from external sources

Functional overview from UniProt



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Report pages combine data from varied sources. Summary data provides quick orientation on what is ‘known’.

Gene: **PPARG** *H. sapiens*

primaryIdentifier: ENSG00000132170 secondaryIdentifier: 5468

symbol: PPARG name: peroxisome proliferator-activated receptor gamma

organism: Homo sapiens mapLocation: 3p25

ncbiGeneNumber: 5468

synonyms: NR1C3, PPARG1, CIMT1, PPARGgamma, PPARG2, GLM1

OMIM Genes UCSC Genes

Literature

1153 publications

firstAuthor	title
Saxena R	Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels.
Zeggini E	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes.
Zeggini E	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes.
Scott LJ	A genome-wide association study of type 2 diabetes in Finns detects multiple
Voight BF	large-scale

Disease

5 diseases

identifier	name
604367	LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 3; FPLD3
125853	DIABETES MELLITUS, NONINSULIN-DEPENDENT; NIDDM
609338	CAROTID INTIMAL MEDIAL THICKNESS 1
137800	GLIOMA SUSCEPTIBILITY 1; GLM1 GLIOMA OF BRAIN, FAMILIAL, INCLUDED
601665	OBESITY LEANNESS, INCLUDED

Receptor that binds peroxisome proliferator-activated receptor ligands and controls the peroxisome homeostasis.

Function overview UniProt

Links to contextual information from external sources

Summary information on Diseases, Homologues, Interaction, Literature, Pathways, GO annotation, and more

Shows which this Gene

Show Physical Interactions | Show Genetic Interactions | Show All Interactions

Interaction Type: — Physical — Genetic

Powered by Cytoscape Web

Report pages draw together additional data to support inference and decision making.

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Evaluate Individual Sequence Variations

- SNPs and Consequence predictions from dbSNP and Ensembl
- Template Searches
 - SNP to GWAS results
 - Region or SNPs to nearest Gene(s)
- List analysis & enrichment
- Find features within chromosome intervals
- *‘Novel’ SNP upload & Consequence prediction*



Evaluate Individual Sequence Variations

Overlap features search from a new list of Genomic Regions

Select the organism and feature types to create and either enter in a list of genomic regions or upload genomic regions from a file. A search will be performed for all the genomic regions in your list.

- Genome regions in the following formats are accepted:
 - **chromosome:start..end**, e.g. *2L:11334..12296*
 - **chromosome:start-end**, e.g. *2R:5866746-5868284* or *chr11:14646344-14667746*
 - **tab delimited**
- Both **base coordinate** (e.g. BLAST, GFF/GFF3) and **interbase coordinate** (e.g. UCSC BED, Chado) systems are supported, users need to explicitly select one. By default, the base coordinate is selected.
- Each genome region needs to take a **new line**.

1. Select Organism:

2. ☒ Select Feature Types:

☒ CDS
☒ Exon
☒ Gene

☒ GeneFlankingRegion
☒ MRNA
☒ SNP

☒ Transcript

3. Type/Paste in genomic regions in ☒ base coordinate ☐ interbase coordinate
([click to see an example](#))

```
chr3:12328767-12475955  
chr21:43841727-43841927  
chrX:99883567-99895088|
```

or Upload genomic regions from a .txt file...



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Summary



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Beyond Metabolic Diseases

Data coverage is not limited to Metabolic Diseases

- All data in metabolicMine are freely available

What does this mean for you?

- Data can be easily embedded into your own web pages!
 - Present up-to-date data sets and information relevant to your genes
 - No overhead of processing and integrating multiple data sources
 - Less updating and storage of data held locally



Gene: **CARTPT**
(**ENSG00000164326**)

Add to New List

Location:	5:71014990-71016875
Length:	1,886
Map Location:	5q13.2
Name:	CART prepropeptide
Ncbi Gene Number:	9,607
Primary Identifier:	ENSG00000164326
Secondary Identifier:	9,607
Summary:	This gene encodes a secreted protein which is processed by prohormone/proprotein convertases to produce smaller, biologically active peptides. Expression of the transcript for this gene is regulated by certain drugs such as cocaine, and the encoded protein is thought to be involved in the regulation of appetite and stress. Mutations in this gene are associated with susceptibility to obesity.
Chromosome:	5
Organism:	Homo sapiens

Embed Gene
summary
information

Homologues

- [Cartpt](#) *Mus musculus*
- [Cartpt](#) *Rattus norvegicus*

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Gene: **CARTPT**
([ENSG00000164326](#))

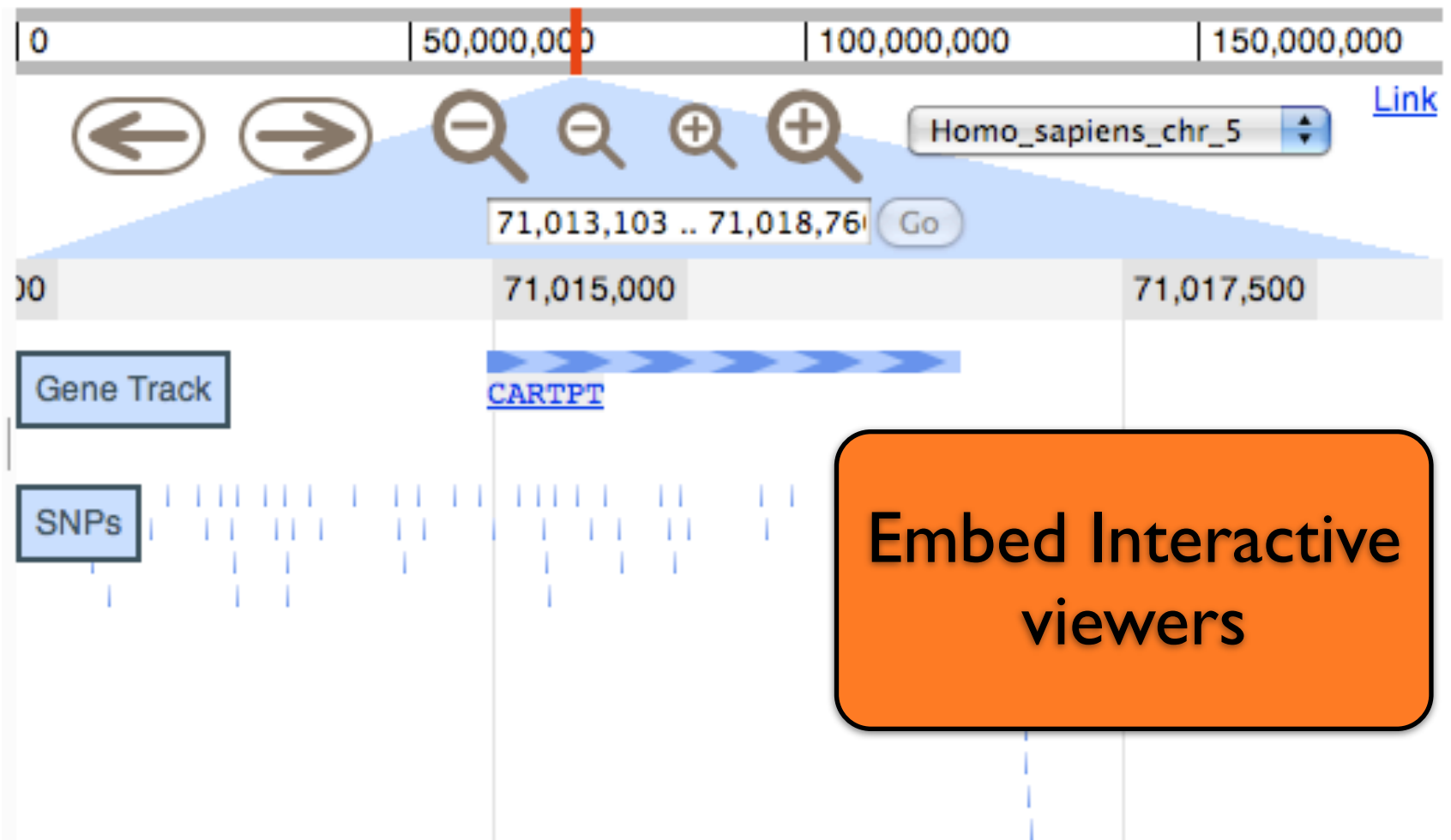
Add to New List

Location: 5:71,014,000-71,016,875

Available Tracks:
(Drag → to view)

DNA

mRNA Track



Homologues

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- [Cartpt](#) *Rattus norvegicus*

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Gene: **CARTPT**
(**ENSG00000164326**)

Add to

New List

Location

Available Length (Drag)

Map Location

DNA

Name mRNA

Ncbi Gene Number

Primary Identifier

Secondary Identifier

Summary

Chromosome

Organism

Homo

• [C](#)

• [Cartpt](#) *Rattus norvegicus*

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Next

	Gene primaryIdentifier	Gene symbol	ontologyTerm identifier	ontologyTerm name	ontologyTerm namespace
1	ENSG00000164326	CARTPT	GO:0000186	activation of MAPKK activity	biological process
2	ENSG00000164326	CARTPT	GO:0001678	cellular glucose homeostasis	biological process
3	ENSG00000164326	CARTPT	GO:0003674	molecular function	molecular function
4	ENSG00000164326	CARTPT	GO:0005576	extracellular region	
5	ENSG00000164326	CARTPT	GO:0005615	extracellular space	
6	ENSG00000164326	CARTPT	GO:0005615	extracellular space	
7	ENSG00000164326	CARTPT	GO:0007165	signal transduction	
8	ENSG00000164326	CARTPT	GO:0007186	G-protein coupled receptor protein signaling pathway	
9	ENSG00000164326	CARTPT	GO:0007218	neuropeptide signaling pathway	biological process
10	ENSG00000164326	CARTPT	GO:0007267	cell-cell signaling	biological process

Export as CSV file

Export as TSV file

View in Mine

Show remaining rows

Load 10 more rows

Embed Template Searches



How To Access the Data

- Through a Java Script library allowing embedding into your own web pages
- Through web services using Perl, Java & Python API

To find out more, visit:

API:	metabolicmine.org/beta/api.do
Embedding tutorial:	intermine.org/imbedding/
Test site:	squirrel.flymine.org/neurogenes/



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Acknowledgments

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MetabolicMine just about to start...

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