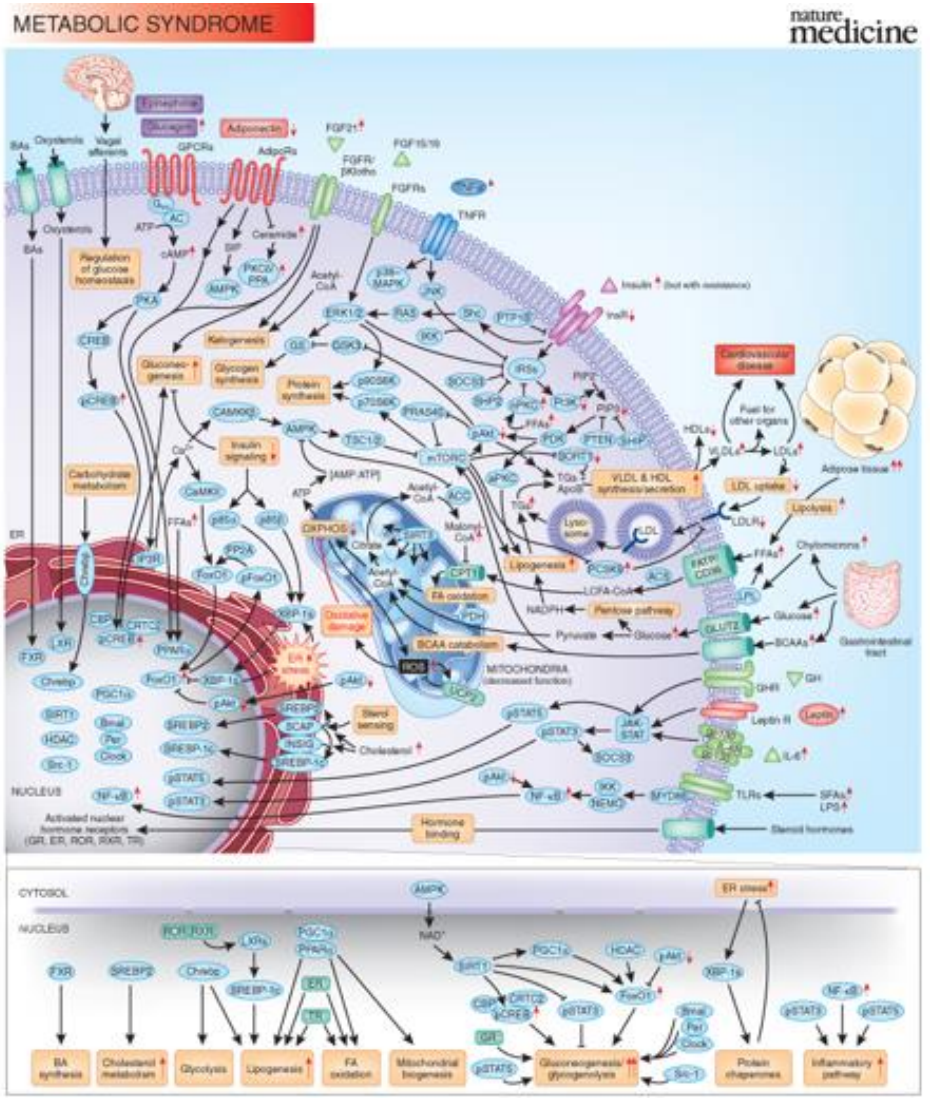
BIOINFORMÁTICA

Otros datos integrativos

Para entender los genomas se necesita información de:

- Vías metabólicas
- Vías regulatorias
- Filogenia



Estudio genético: Alzheimer

¿Cuál es el riesgo de enfermar en caso de encontrar alguna mutación genética específica?

Información en
1×10^9 b
de secuencia de ADN

Secuenciar a una persona

Secuencia de ADN

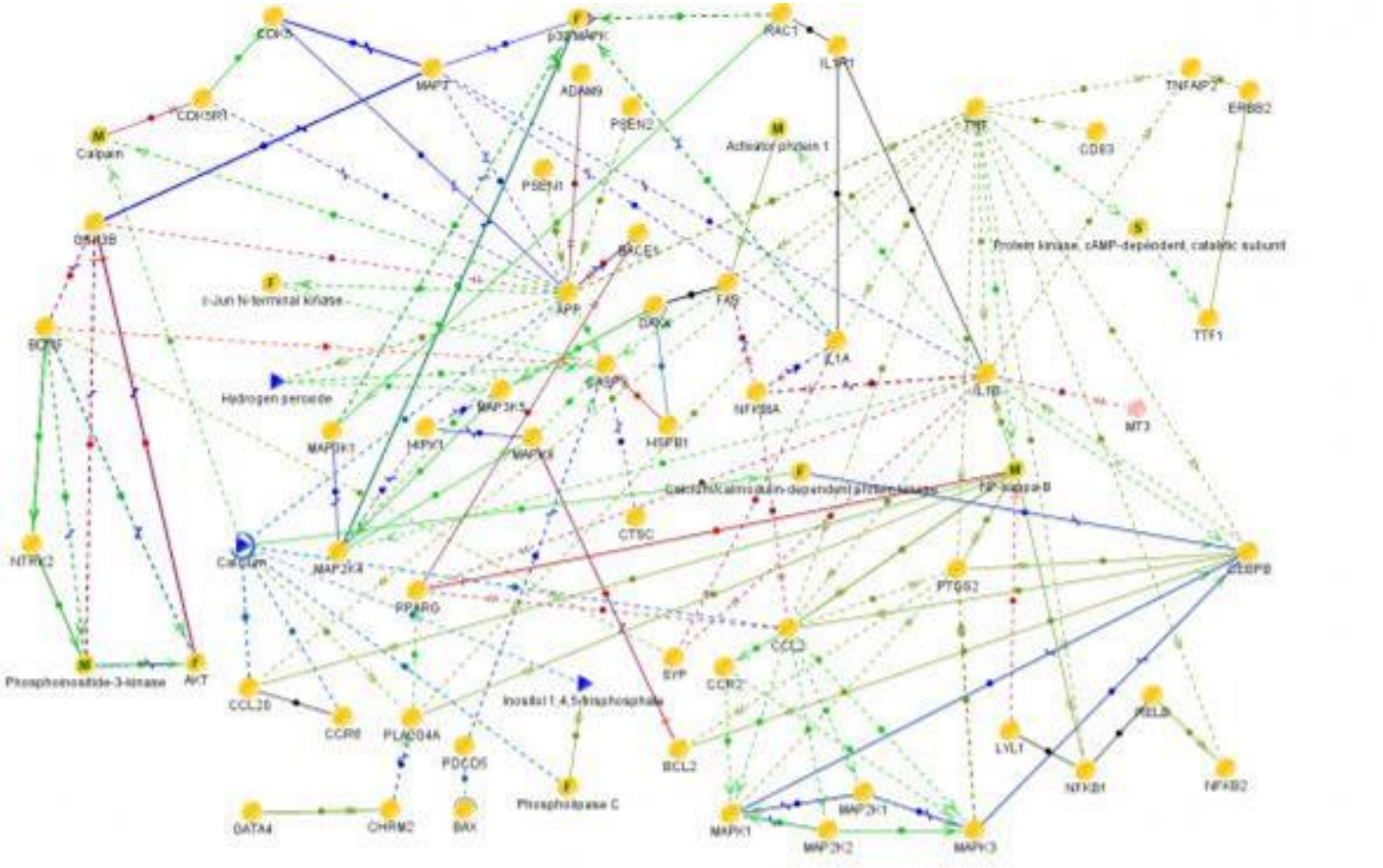
Regulación epigenética

Expresión génica

¿Qué puede hacer la medicina?

Análisis de redes moleculares

usando herramientas bioinformáticas



La clave: la colaboración

Entre los tres navegadores de genoma principales:

- Ensembl, UCSC y NCBI
  - Ensembl Genome Browser
    http://www.ensembl.org/
  - NCBI Map Viewer
    http://www.ncbi.nlm.nih.gov/mapview/
  - UCSC Genome Browser
    http://genome.ucsc.edu/

Navegadores de genoma

- Navegar genes en su contexto genómico
- Ver características dentro y alrededor de un gen específico
- Investigar la organización del genoma y explorar grandes regiones de cromosomas
- Buscar y obtener información a escala de gen y de genoma
- Comparar genomas

Alzheimer desde la genética

El gen APOE, en el cromosoma 19, consiste de:

- Cuatro exones
- Tres intrones
- 3597 pares de bases

Entre sus funciones, da instrucciones para producir la proteína apolipoproteína E.

http://ghr.nlm.nih.gov/gene/APOE

Lo conocido de APOE en 2015

El gen APOE es polimórfico, con tres mayores alelos:

- APOE E2 (7%) - enlaza pobremente receptores de superficie. Se asocia con riesgo para arteriosclerosis.
- APOE E3 (79%) - el «neutral»
- APOE E4 (14%) - asociado a altos niveles de vitamina D, Alzheimer, apnea del sueño, resultado desfavorable tras una lesión cerebral traumática, acortamiento acelerado de telómeros y enfermedad cerebrovascular isquémica.

UCSC Genome browser

http://genome.ucsc.edu/

UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Session - FAQ - Help

Genome Browser
Ebola
Blat
Table Browser
Gene Sorter
In Silico PCR
Genome Graphs
Galaxy
VisiGene
Utilities
Downloads
Release Log
Custom Tracks
Cancer Browser
Microbial Genomes
ENCODE
Neandertal

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 Neandertal project. 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. VisiGene 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 and the Center for Biomolecular Science and Engineering (CBSE) 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 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.

DONATE NOW

News 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.

12 February 2015 - Blat Your Assembly Hub

Assembly data hubs, track hubs that allow researchers to annotate genomes that are not in the UCSC Genome Browser, can now use blat to quickly find DNA and protein sequences in their unique assemblies.

While running remote blat servers with the gfServer utility, you can now add lines to your assembly hub's genomes.txt file to inform the Browser where to send blat searches. Or by downloading and installing a virtual machine Genome Browser In a Box (GBiB), you can alternatively locally run gfServers, preinstalled on GBiB, all from your laptop. For more information see the Assembly Hub Wiki and the new Quick Start Guide to Assembly Hubs.

11 February 2015 - dbSNP 142 Available for hg19 and hg38

Gene Sorter, APOE

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UCSC Human Gene Sorter

genome Human | assembly Feb. 2009 (GRCh37/hg19) | search APOE | Go!

sort by Expression (GNF Atlas2) | configure | filter (now off) | display 50 | output sequence | text

#	Name	VisiGene	fetal brain	whole brain	amygdala	thymus	bone marrow	PB.CD4+ T cells	skin	adipocyte	pancreatic islets	heart	lung	kidney	liver	ovary	testis	BLASTP E-Value	Genome Position	Description
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2	APOC1P1	n/a																n/a	chr19:45,432,351	Homo sapiens apolipoprotein C-I pseudogene 1 (APOC1P1), transcript variant 1, nor
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4	EPHX1	176110																n/a	chr1:226,015,529	Homo sapiens epoxide hydrolase 1, microsomal (xenobiotic) (EPHX1), transcript vari
5	DDAH1	166924																n/a	chr1:85,857,528	Homo sapiens dimethylarginine dimethylaminohydrolase 1 (DDAH1), transcript variar
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7	EFHD1	n/a																n/a	chr2:233,522,849	Homo sapiens EF-hand domain family, member D1 (EFHD1), transcript variant 1, mR
8	FXYD1	176440																n/a	chr19:35,631,843	Homo sapiens FXYD domain containing ion transport regulator 1 (FXYD1), transcript
9	LARP6	172487																n/a	chr15:71,135,193	Homo sapiens La ribonucleoprotein domain family, member 6 (LARP6), transcript var
10	PCDHGC5	150326																n/a	chr5:140,880,678	Homo sapiens protocadherin gamma subfamily C, 5 (PCDHGC5), transcript variant 1
11	PDE2A	n/a																n/a	chr11:72,336,340	Homo sapiens phosphodiesterase 2A, cGMP-stimulated (PDE2A), transcript variant
12	TSPAN7	67458																n/a	chrX:38,484,451	Homo sapiens tetraspanin 7 (TSPAN7), mRNA.
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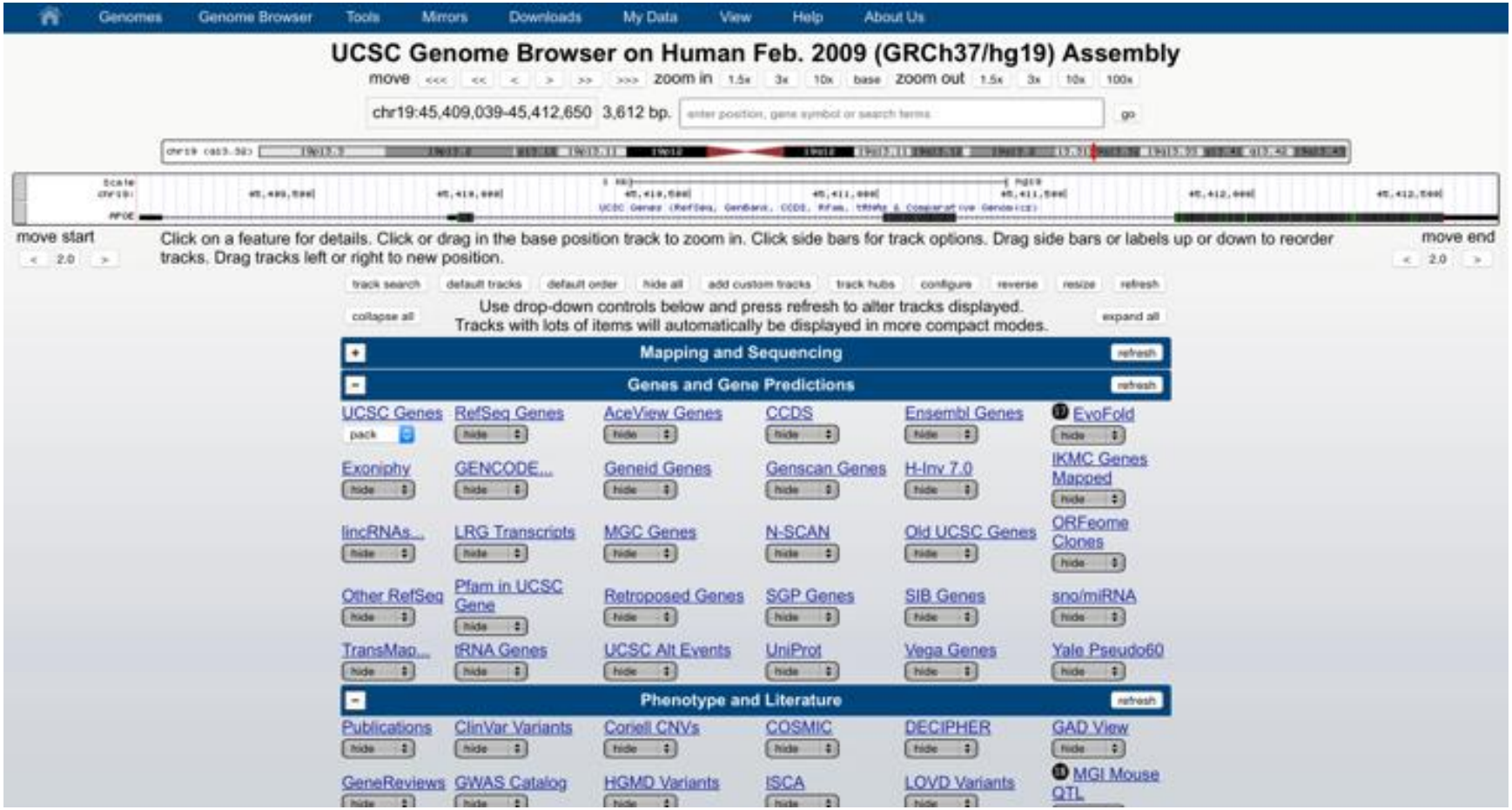
Genomes
Genome Browser
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Mirrors
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Help
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UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
zoom in
zoom out
chr19:45,409,039-45,412,650 3,612 bp.
go
move start
move end
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.
track search
default tracks
default order
hide all
add custom tracks
track hubs
configure
reverse
resize
refresh
collapse all
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.
expand all
Mapping and Sequencing
Genes and Gene Predictions
UCSC Genes
RefSeq Genes
AceView Genes
CCDS
Ensembl Genes
EvoFold
Exoniphy
GENCODE...
Geneid Genes
Genscan Genes
H-Inv 7.0
IKMC Genes Mapped
lincRNAs...
LRG Transcripts
MGC Genes
N-SCAN
Old UCSC Genes
ORFeome Clones
Other RefSeq
Pfam in UCSC Gene
Retroposed Genes
SGP Genes
SIB Genes
sno/miRNA
TransMap...
tRNA Genes
UCSC Alt Events
UniProt
Vega Genes
Yale Pseudo60
Phenotype and Literature
Publications
ClinVar Variants
Coriell CNVs
COSMIC
DECIPHER
GAD View
GeneReviews
GWAS Catalog
HGMD Variants
ISCA
LOVD Variants
MGI Mouse QTL

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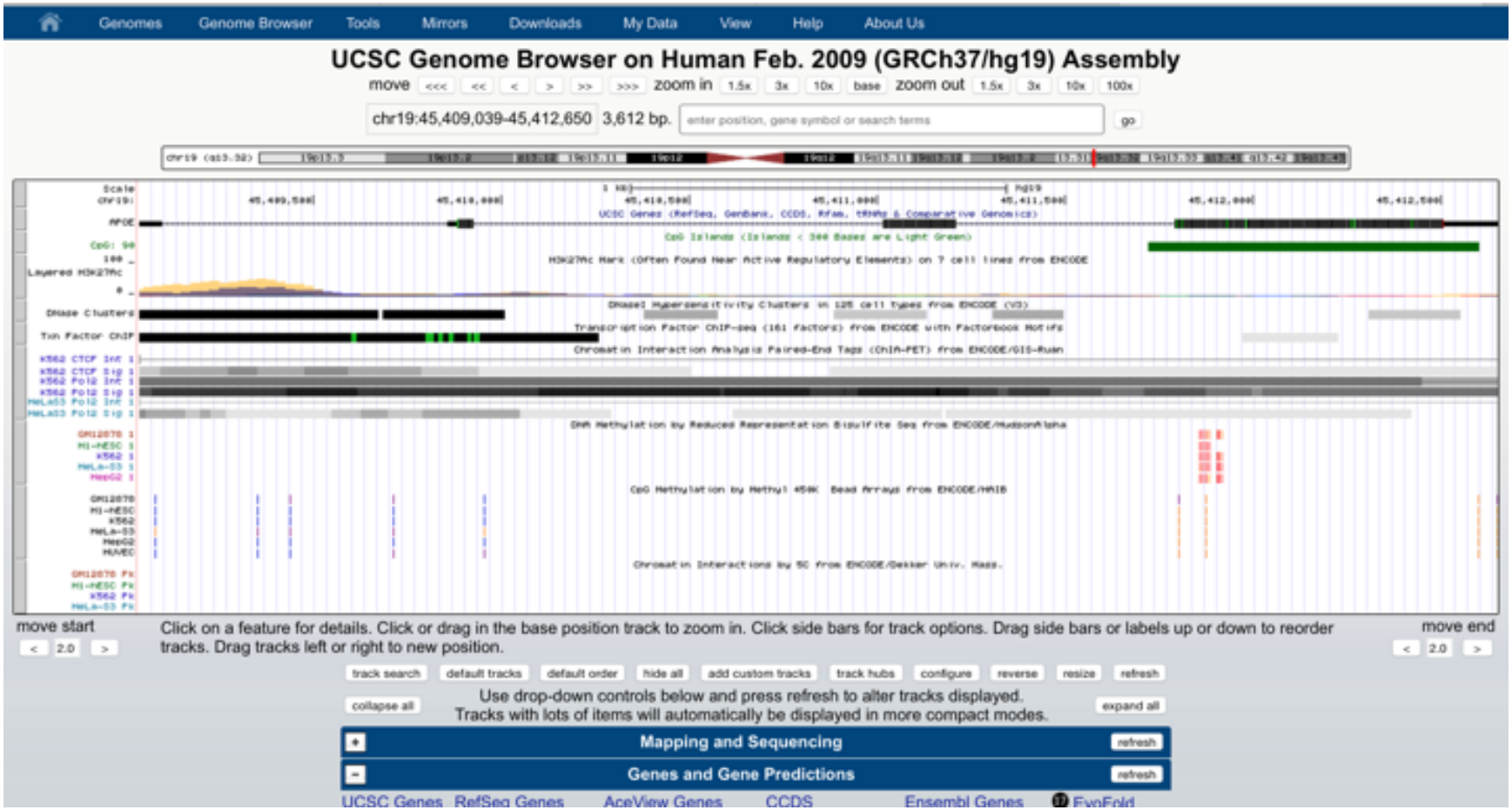
Allen Brain
Burge RNA-seq
CSHL Small RNA-seq
ENC Exon Array...
ENC ProtGeno...
ENC RNA-seq...
GIS RNA PET
GNF Atlas 2
Illumina WG-6
PeptideAtlas...
qPCR Primers
RIKEN CAGE Loc
Sestan Brain
Regulation
ENCODE Regulation...
CD34 DnaseI
CpG Islands...
ENC Chromatin...
ENC DNA Methyl...
ENC DNase/FAIRE...
ENC Histone...
ENC RNA Binding...
ENC TF Binding...
FSU Repli-chip
Genome Segments
NKI Nuc Lamina...
ORegAnno
Stanf Nucleosome
SUNY SwitchGear
SwitchGear TSS
TFBS Conserved
TS miRNA sites
UCSF Brain Methyl
UMMS Brain Hist
UW Repli-seq
Vista Enhancers
Comparative Genomics
Conservation
Cons 46-Way
Cons Indels MmCf
Evo Cpg
GERP
phastBias gBGC
Primate Chain/Net
Placental Chain/Net
Vertebrate Chain/Net
Neandertal Assembly and Analysis
Denisova Assembly and Analysis
Variation
Common
1000G Ph1 Accsbl
1000G Ph1 Vars
All SNPs(138)
All SNPs(141)
All SNPs(142)

Regulación epigenética del gen



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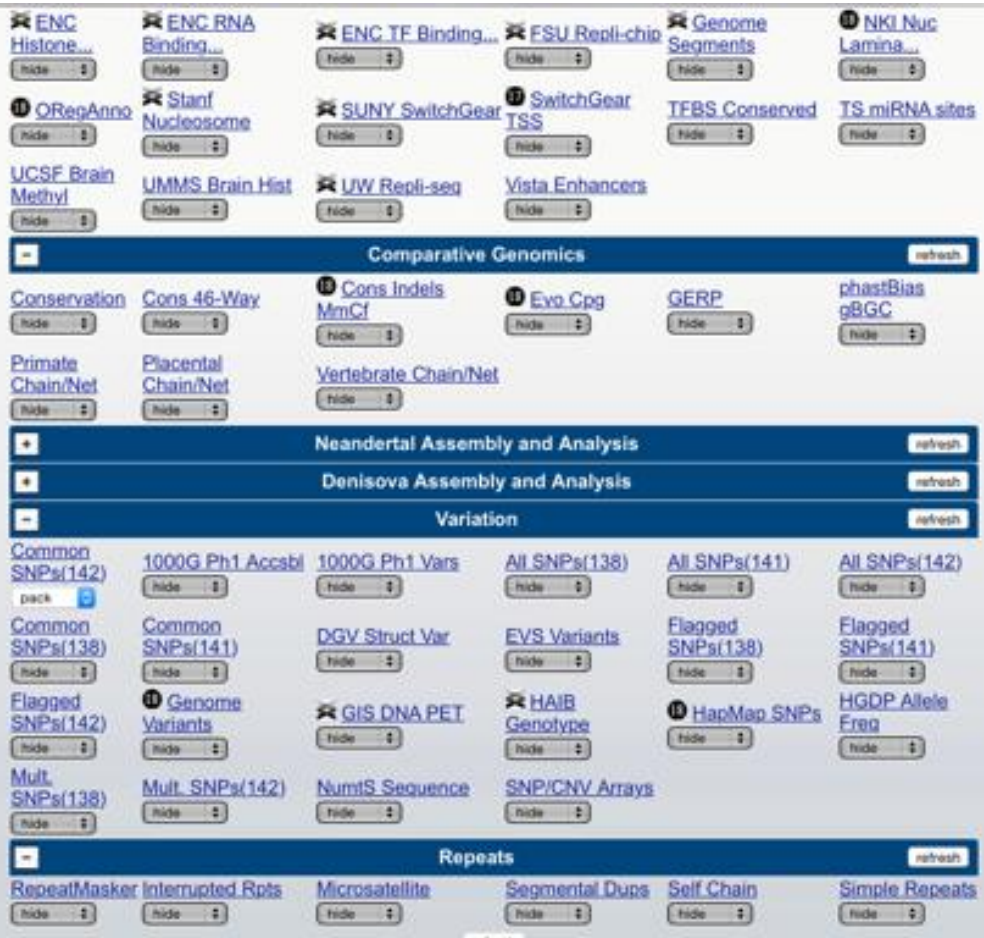
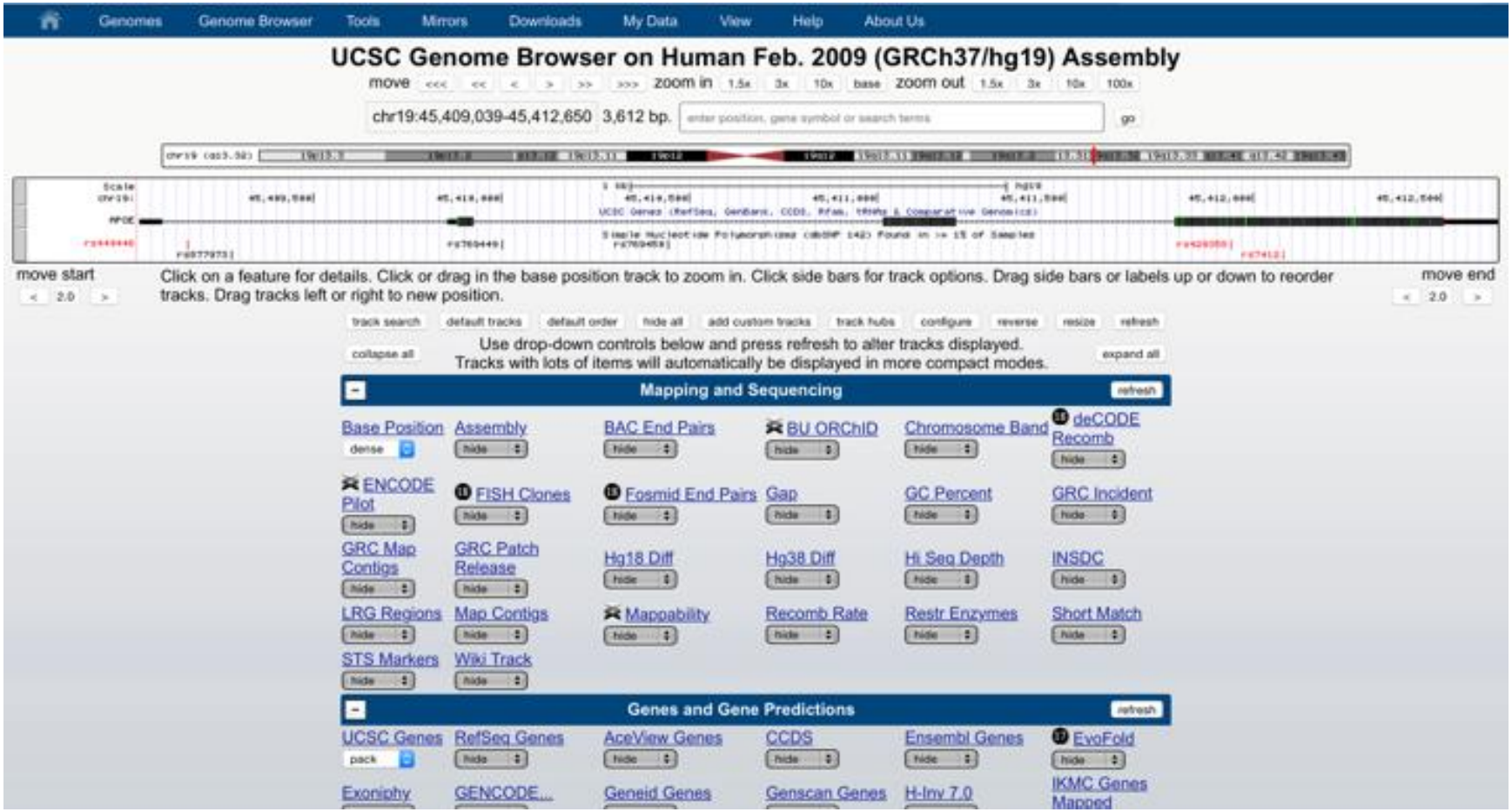

ENC Histone...
ENC RNA Binding...
ENC TF Binding...
FSU Repli-chip
Genome Segments
NKI Nuc Lamina...
ORegAnno
Stanf Nucleosome
SUNY SwitchGear
SwitchGear TSS
TFBS Conserved
TS miRNA sites
UCSF Brain Methyl
UMMS Brain Hist
UW Repli-seq
Vista Enhancers
Comparative Genomics
refresh
Conservation
Cons 46-Way
Cons Indels MmCf
Evo Cpg
GERP
phastBias gBGC
Primate Chain/Net
Placental Chain/Net
Vertebrate Chain/Net
Neandertal Assembly and Analysis
Denisova Assembly and Analysis
Variation
Common SNPs(142)
pack
1000G Ph1 Accsbl
1000G Ph1 Vars
All SNPs(138)
All SNPs(141)
All SNPs(142)
Common SNPs(138)
Common SNPs(141)
DGV Struct Var
EVS Variants
Flagged SNPs(138)
Flagged SNPs(141)
Flagged SNPs(142)
Genome Variants
GIS DNA PET
HAIB Genotype
HapMap SNPs
HGDP Allele Freq
Mult. SNPs(138)
Mult. SNPs(142)
NumtS Sequence
SNP/CNV Arrays
Repeats
RepeatMasker
Interrupted Rpts
Microsatellite
Segmental Dups
Self Chain
Simple Repeats
hide

Variaciones comunes del gen:



Alzheimer en 2023

~15 años atrás	Hoy	Futuro
¿Riesgo?	Modificación de factores de riesgo ambientales Determinación de riesgo genético	Modificación de factores de riesgo genéticos Restablecimiento de proteínas afectadas, vías metabólicas, etc.

Información altamente cambiante

Gen ATG - dicta el aminoácido metionina - su falla puede desembocar en aterosclerosis.[1]

20 genes vinculados a la autofagia (**AuTophaGy related)**[2]

33 genes relacionados a la autofagia y la construcción de vesículas autofagosomas y de focalización citoplasma a vacuola (Cvt).[3]

62 genes ligados a autofagia, construcción de vesículas autofagosomas, focalización citoplasma a vacuola (Cvt) y APOE4 se relaciona a incrementar riesgo de demencia por problemas en el metabolismo de la Colina y ciertos lípidos.[4]

Metaanálisis de variaciones asociadas de APOE E4, Alzheimer y demencias en Latinoamérica.[5] Protección asociada a la apolipoproteína E4 contra infecciones entéricas pediátricas como ventaja de supervivencia en poblaciones preindustriales.[6]

1. http://circres.ahajournals.org/content/104/3/304.full
2. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3442874/
3. http://www.genenames.org/cgi-bin/genefamilies/set/1022
4. https://www.nia.nih.gov/news/study-reveals-how-apoe4-gene-may-increase-risk-dementia
5. https://content.iospress.com/articles/journal-of-alzheimers-disease/jad221167
6. https://pubmed.ncbi.nlm.nih.gov/37125551/

Aplicaciones de la bioinformática

- Análisis de secuencias
- Análisis de expresión de proteínas
- Anotación de genomas
- Análisis de mutaciones en el cáncer
- Biología evolutiva computacional
- Genómica comparativa
- Acoplamiento proteína-proteína
- Predicción de la estructura de las proteínas
- Análisis de expresión génica y de la regulación
- Modelado de sistemas biológicos
- Medición de la biodiversidad

Más información

- Swan, Melanie. (2009). *Next-generation Personal Genomic Studies: Extending Social Intelligence Genomics to Cognitive Performance Genomics in Quantified Creativity and Thinking Fast and Slow hacer más efectivas las presentaciones* científicas. http://www.diygenomics.org/files/AAAI_2013_SWAN.pdf
- Prada, Diddier, MD. (2015). *Bioinformática*. Laboratorio de Epigenética ambiental, Escuela de Salud Pública, Harvard. http://www.hsph.harvard.edu/biostatistics/bioinformatics-computational-biology/
- Jacob, Joachim (2011). *Basic bioinformatics concepts, databases and tools. Module 5. Genome browsers and interpretation of gene lists*. http://www.slideshare.net/joachimjacob/mod5-intro-h12011genomebrowsers?qid=ca415995-3fd3-475b-b929-a7c101253e14&v=default&b=&from_search=4
- Epigenie. *Histone H3K27*. http://epigenie.com/key-epigenetic-players/histone-proteins-and-modifications/histone-h3k27/