



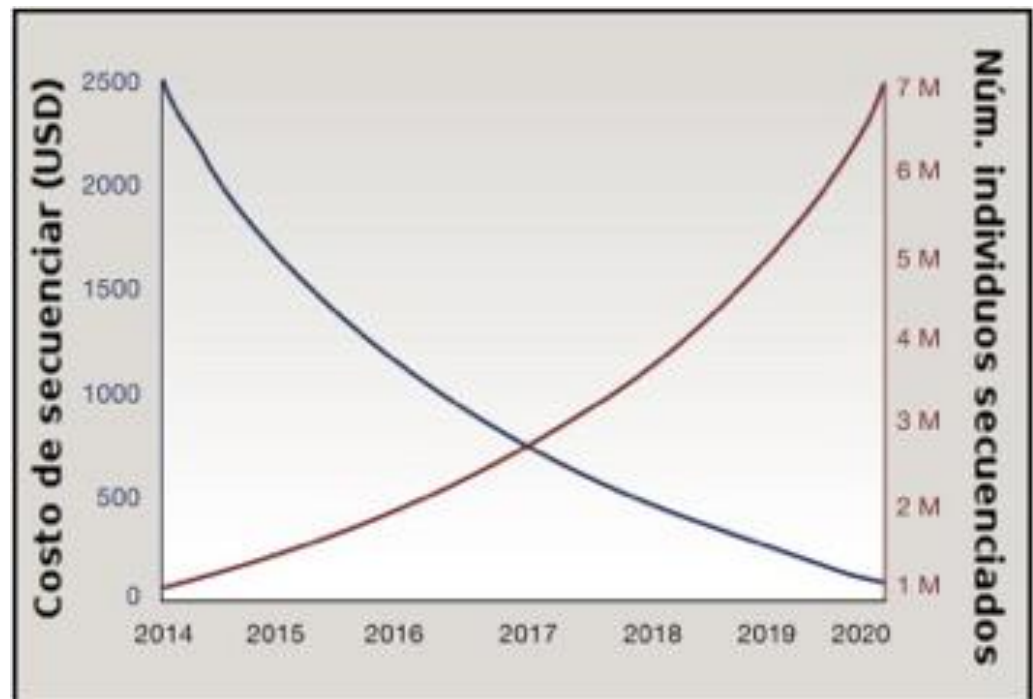
BIOINFORMÁTICA

Información de todo el genoma



Genomas

- Bacteria: 1.6 Mb (1995)
- Eucarionte: 13 Mb (1997)
- Animal: 100 Mb (1998)
- Humano: 3 Gb (2001)



Método de secuenciación de disparo a todo el genoma



DNA genoma



Secuenciar cada fragmento con acercamiento de disparo

GCTTTGAGTTTCCCTGACACCACTT

CCAGTGGTACTGAGACCCAGAGGCTTA

GCTTATTTGCCATTTATTTATTT

Alinear **secuencias** **contiguas**

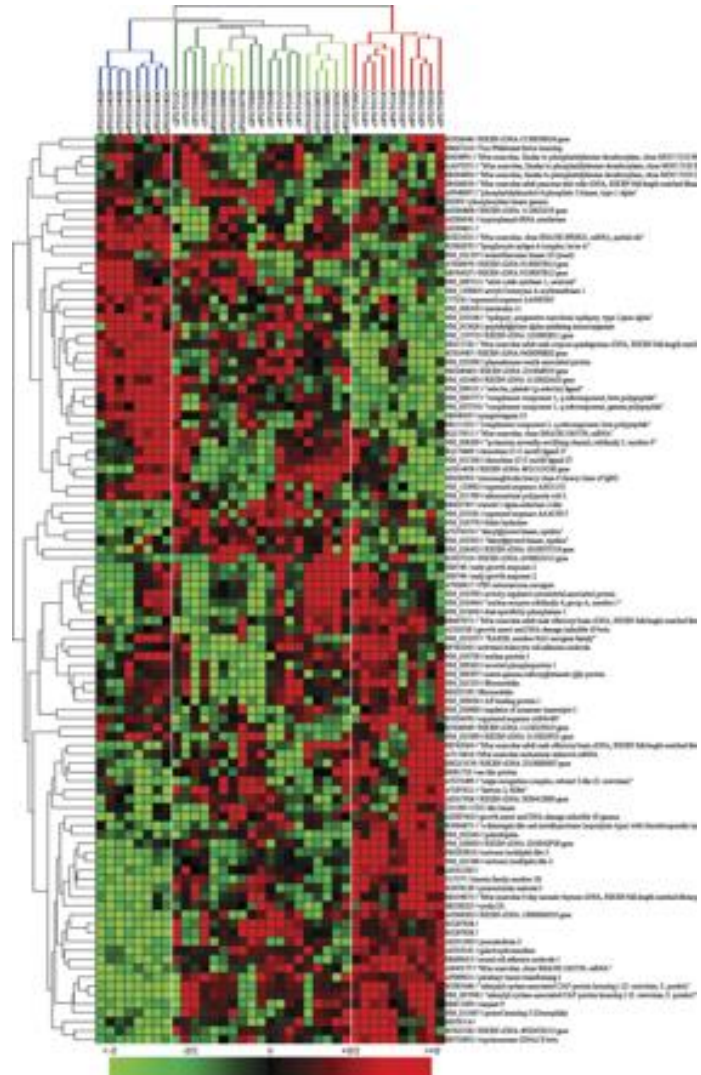
GCTTTGAGTTTCCCTGACACCACTTGGTACTGAGACCCAGAGGCTTATTTGCCATTTATTTATTT

Secuencia generada terminada

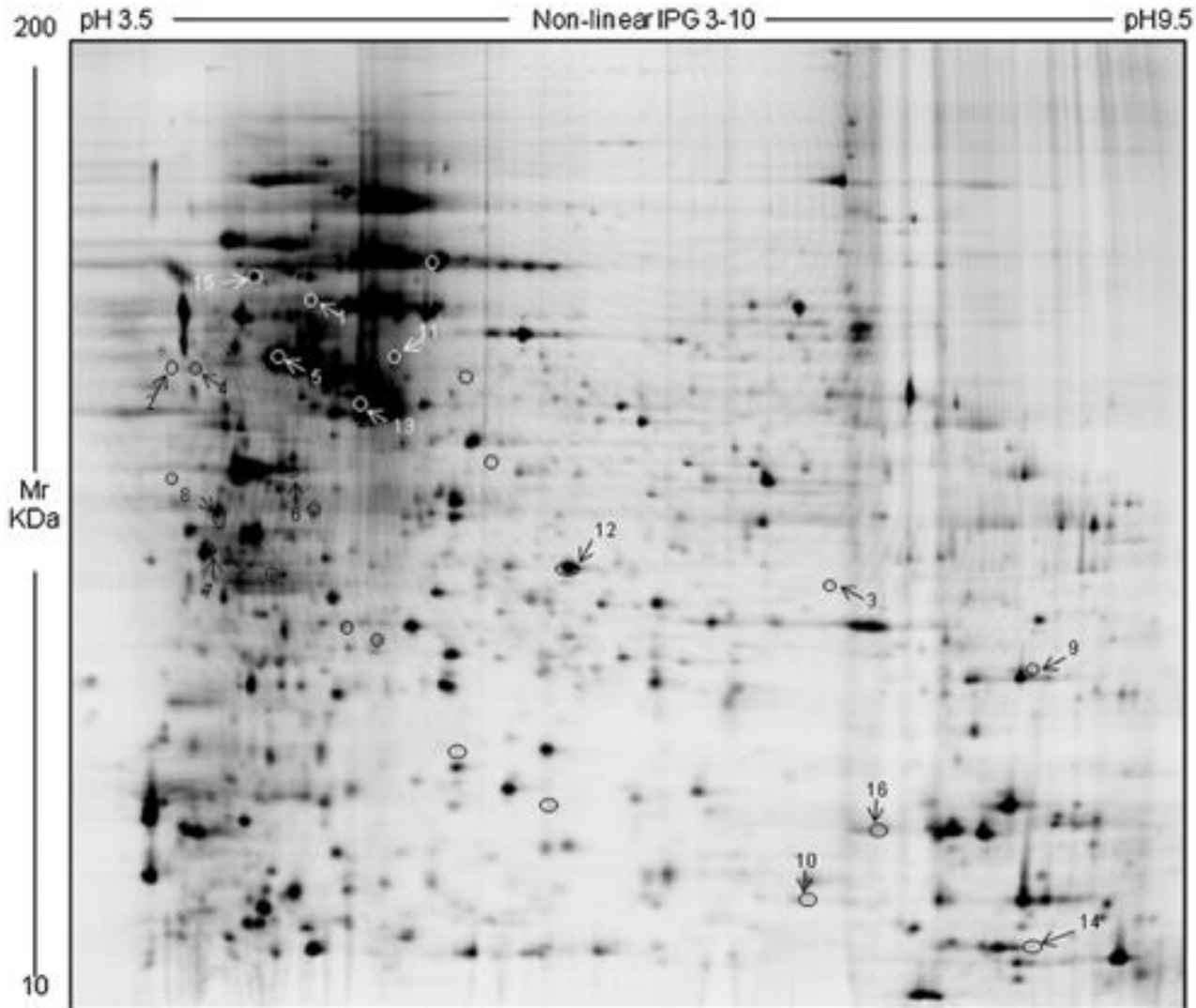
Transcriptoma: bases de datos con la expresión de todos los genes



Heatmap de un
microarreglo de
expresión



Proteoma: bases de datos con la expresión de todas las proteínas

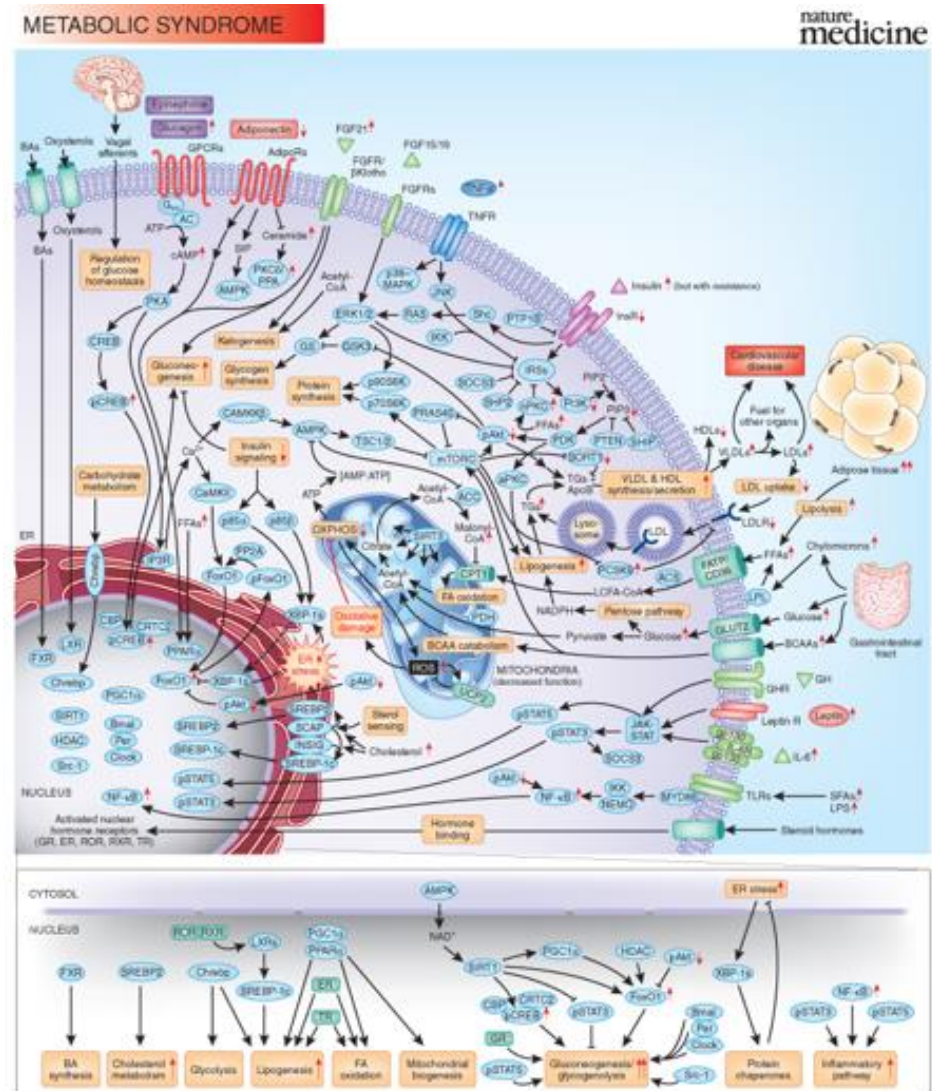


Otros datos integrativos

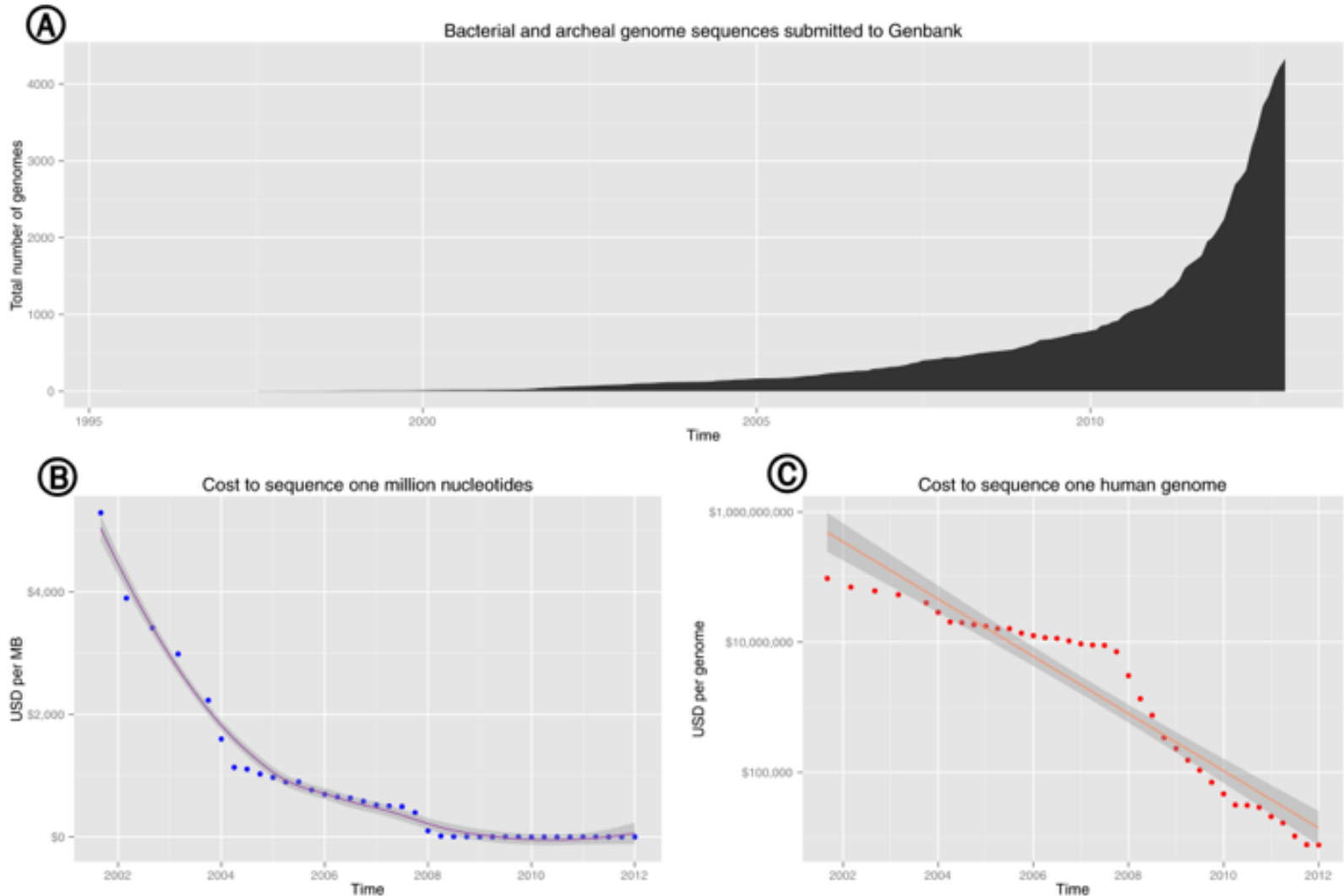


Para entender los genomas se necesita información de:

- Vías metabólicas
- Vías regulatorias
- Filogenia



Crecimiento exponencial de datos asociados al desarrollo de TIC



Ejemplo del equipo empleado: Un cluster típico



Cada nodo tiene *cores* múltiples de CPU:



Cada chasis tiene 16 nodos:



Conectando una cantidad de nodos se obtiene un cluster:



Estudio genético: Alzheimer



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

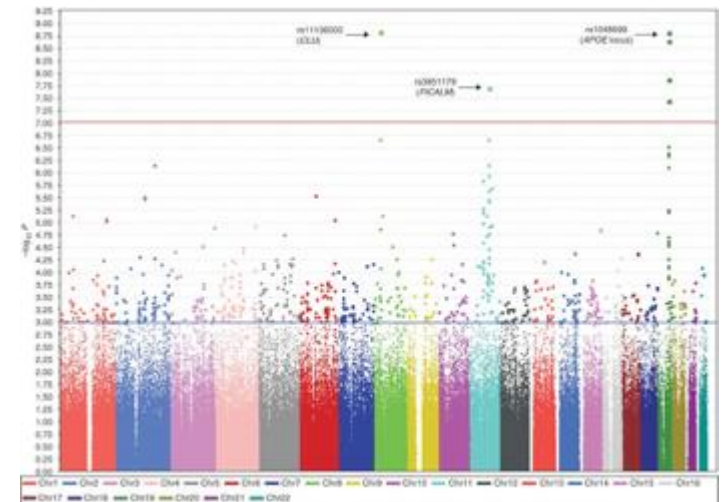
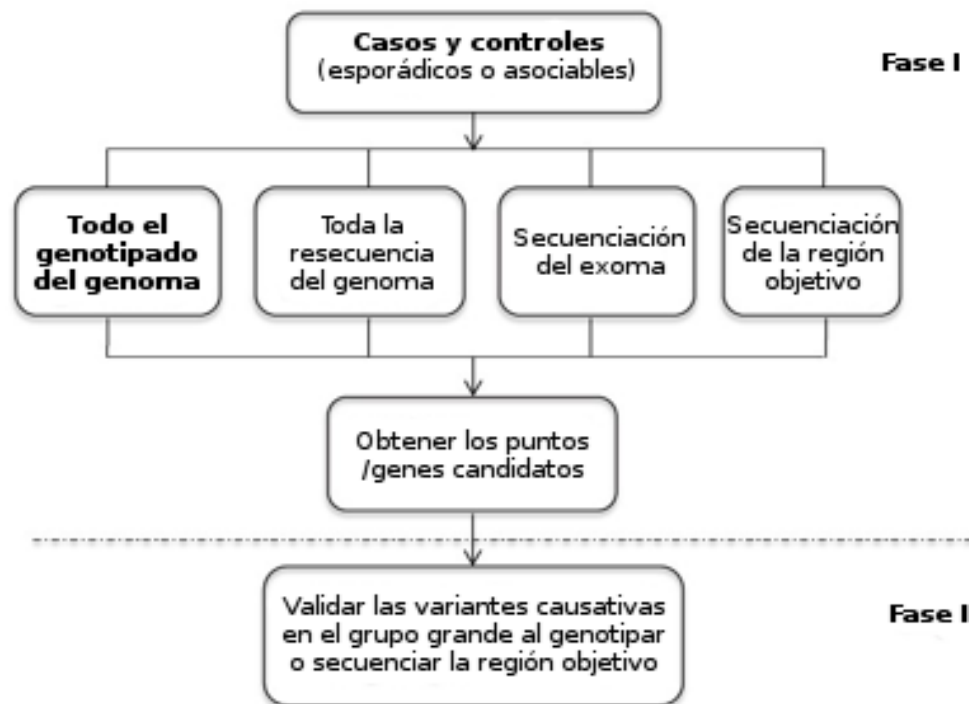
Información en
 1×10^9 b
de secuencia de ADN



Análisis GWAS



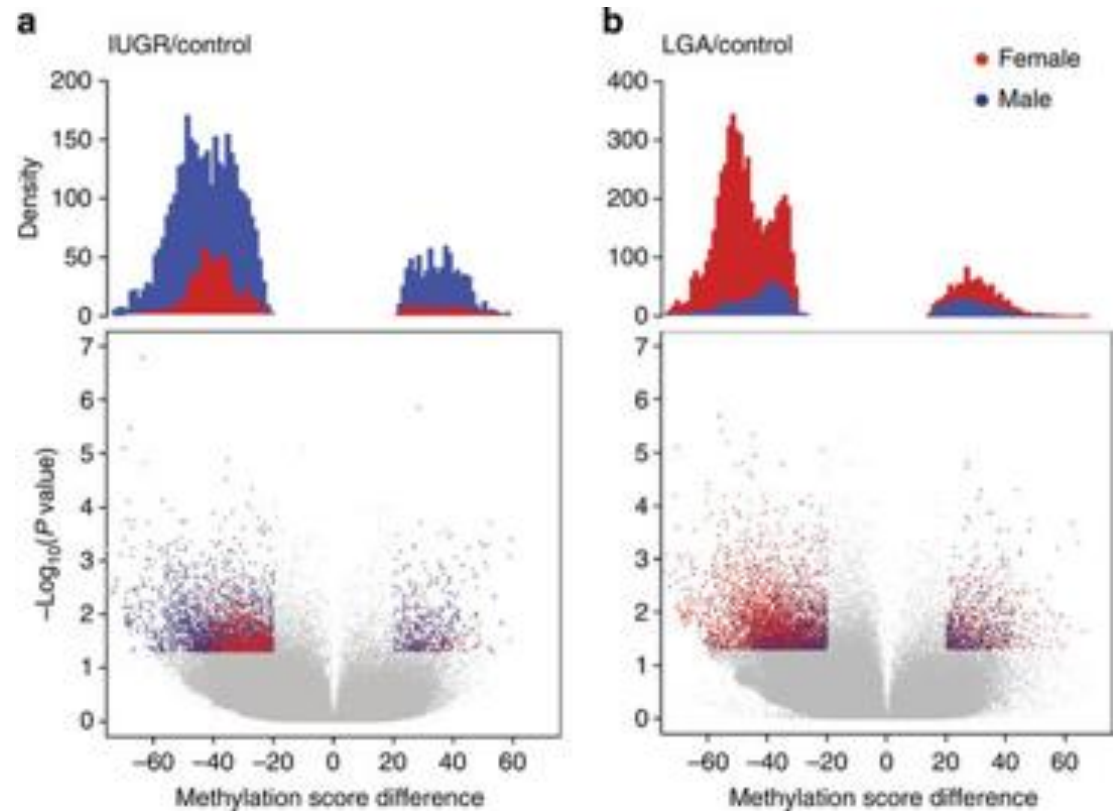
Estudios de asociación amplia genómica usando herramientas bioinformáticas



Análisis GWAS



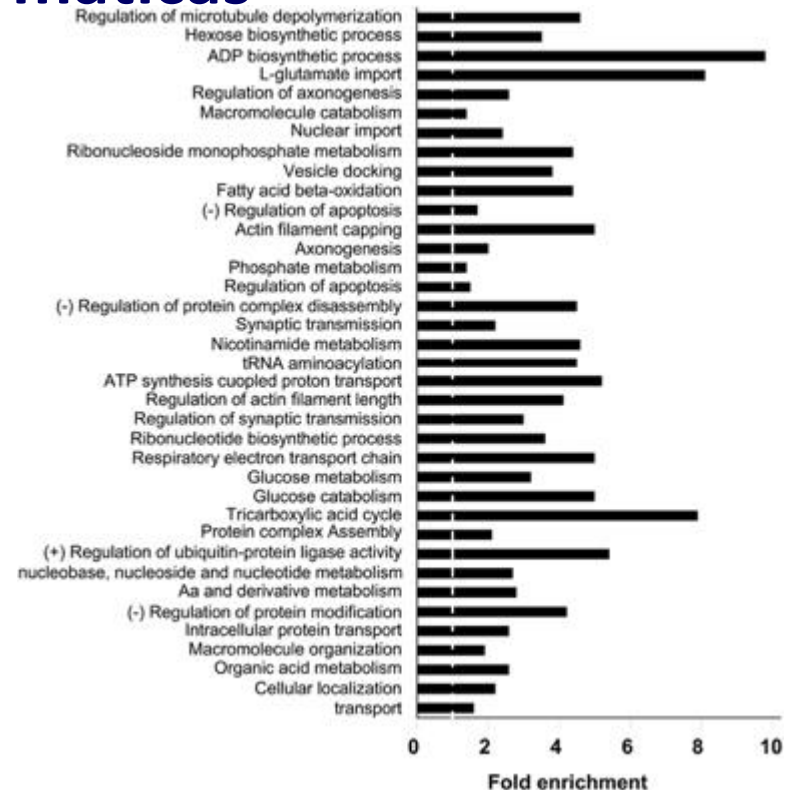
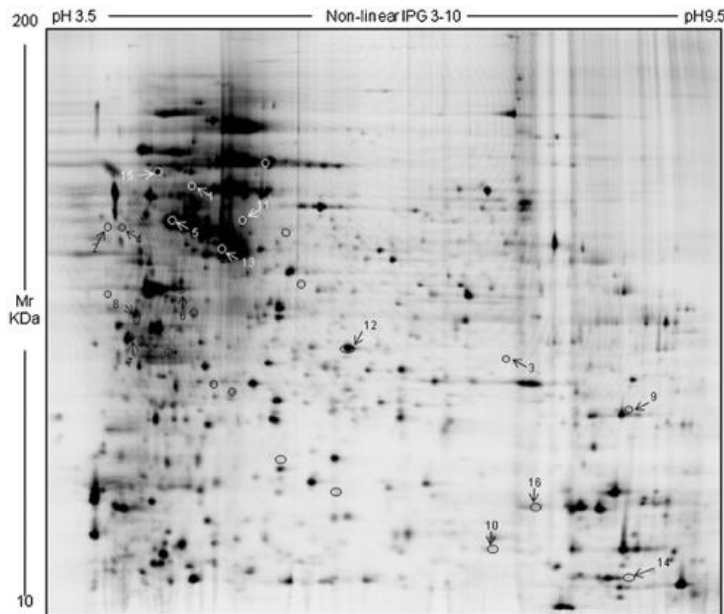
Illumina 450,000 sitios CpG
usando herramientas bioinformáticas



Análisis GWAS



Patrón de expresión de proteínas en regiones específicas usando herramientas bioinformáticas



Secuenciar a una persona



Secuencia de ADN

Regulación
epigenética

Expresión génica

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>

UCSC Genome browser



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

UCSC Genome Bioinformatics

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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 ([GBB](#)) 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](#).

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News

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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 ([GBB](#)), you can alternatively locally run gfServers, preinstalled on GBB, 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