

# Introduction to Biological Databases

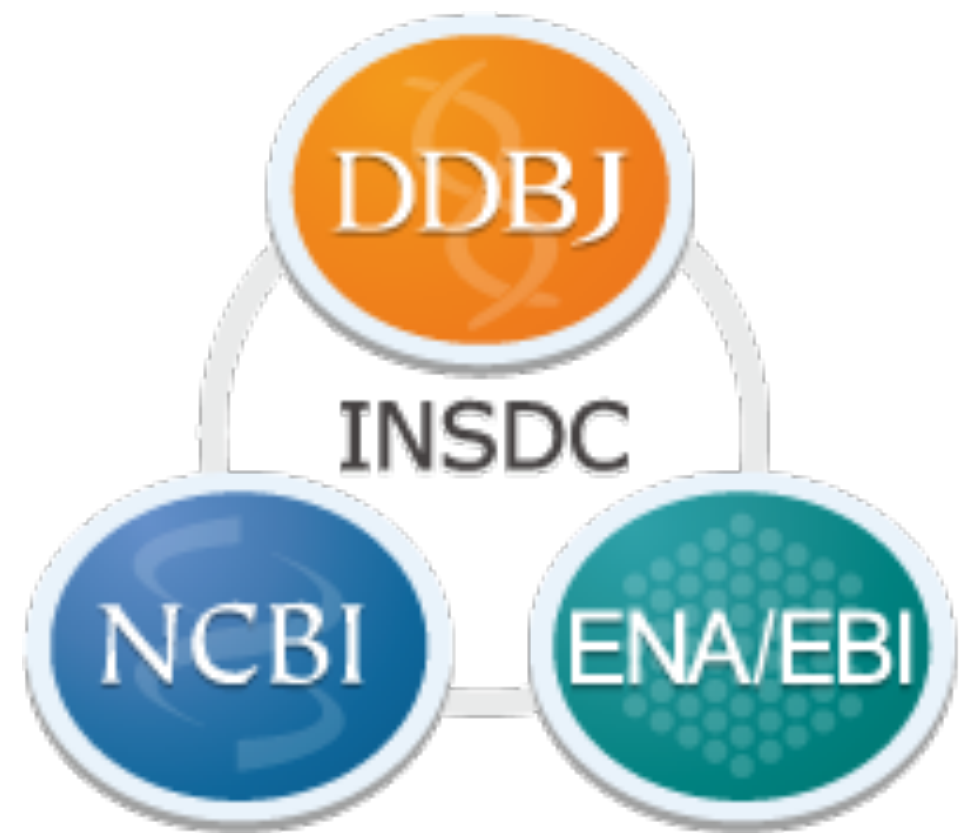
# Biological Databases

a repository of data collected from scientific experiments, published literature, high-throughput technology, or computational analyses

- ▶ host of different research areas
- ▶ provide a single point of access

# International Nucleotide Sequence Database Collaboration

- ▶ A longstanding global initiative
- ▶ Creating a comprehensive collection of public domain nucleotide sequences and metadata
- ▶ Worldwide synchronization of sequence data (submit anywhere; daily updates)
- ▶ Currently include sequence data from >160,000 species



# EMBL-EBI



European Nucleotide Archive, established in 1980 at EMBL in Heidelberg, Germany

- Central database of DNA sequences
- EMBL-EBI established in UK on Wellcome Trust campus 1992; transition of two major bioinformatics services
- Provide a comprehensive range of molecular databases and offer extensive user training programme





In 1984 briefing sessions began on Capitol Hill; Dr. Allan Maxam germinated the idea of the Center

Created in 1988 as part of the National Library of Medicine at NIH

- Establish public databases
- Research in computational biology
- Develop software tools for sequence analysis
- Disseminate biological information





EMBL and Genbank started international cooperation and invited Japan to participate

- trial data loading was started in 1983, and the next year DDBJ as up and running as part of the National Genetics Institute (NGI)
- To operate DDBJ more efficiently, the Center for Information Biology (CIB) was established within the NIG in 1995
- Services include biological database management and various software tools for data analysis (web services and NIG supercomputer)



# Sequence data

- ▶ Contain data from individual organisms, specific categories/functions of sequences, or data generated by specific sequencing technologies. Examples:
  - ▶ Organism-specific
  - ▶ Sequence categories or functions
  - ▶ Data generated by specific sequencing technologies (EST, STS, HTG)

## Mus musculus GRIN1 (Z16) mRNA, complete cds

GenBank: AF146569.1

[FASTA](#) [Graphics](#)

Go to: ☐

LOCUS AF146569 3568 bp mRNA linear ROD 16-SEP-1999  
DEFINITION Mus musculus GRIN1 (Z16) mRNA, complete cds.  
ACCESSION AF146569  
VERSION AF146569.1 GI:5901687  
KEYWORDS .  
SOURCE Mus musculus (house mouse)  
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Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;  
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USA

FEATURES Location/Qualifiers  
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gene 1..3568  
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901 tctgggcagg cagaacgctg gtccttggtg aaaacagaaa cattatcctc aggaaaagaa
```

## Flatfile format for INDSC:

- Header



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901 tctgggcagg cagaacgcgt gtccttggtg aaaacagaaa cattatcctc aggaaaagaa
```

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- Feature Table: details about the sequence

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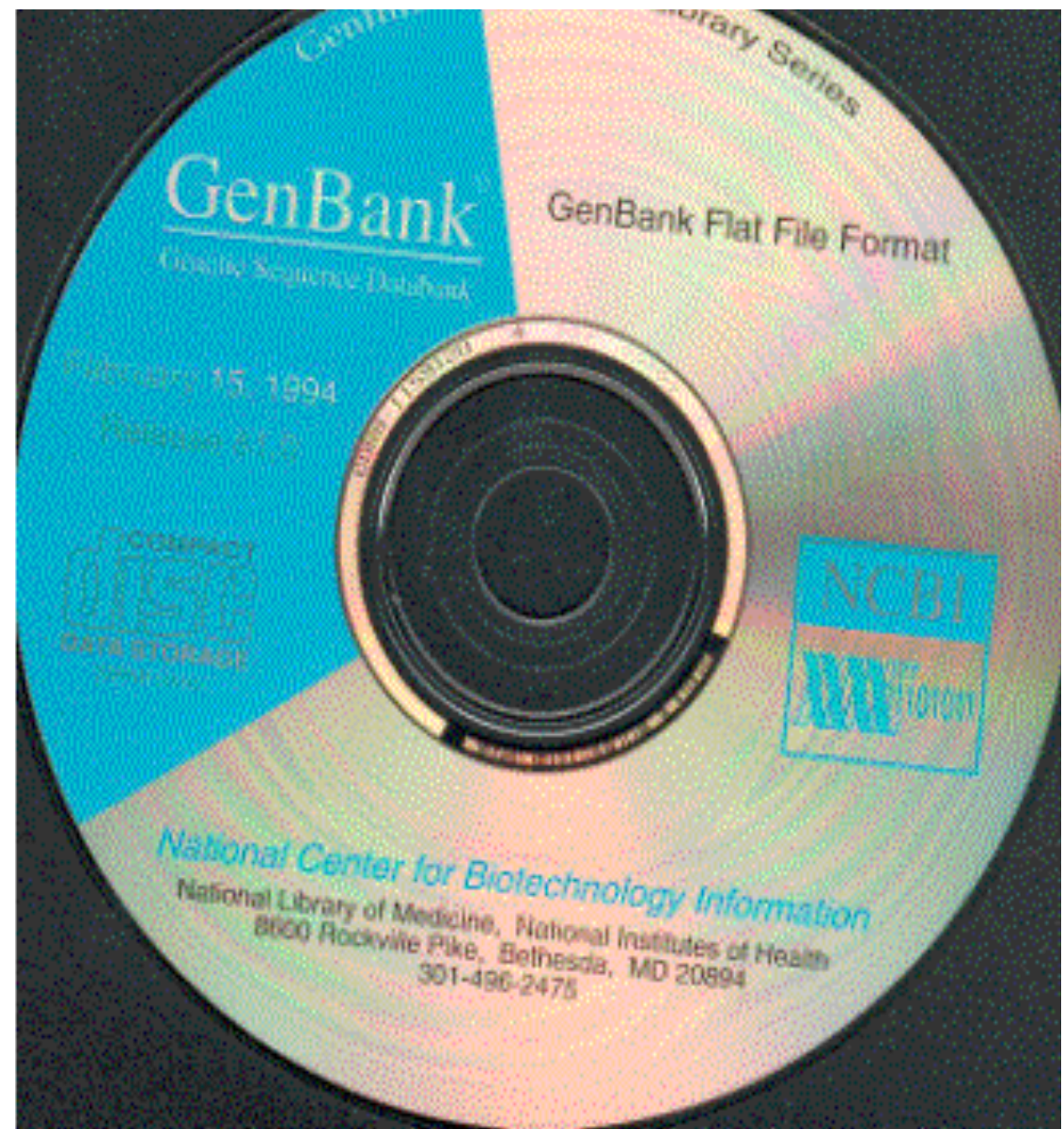
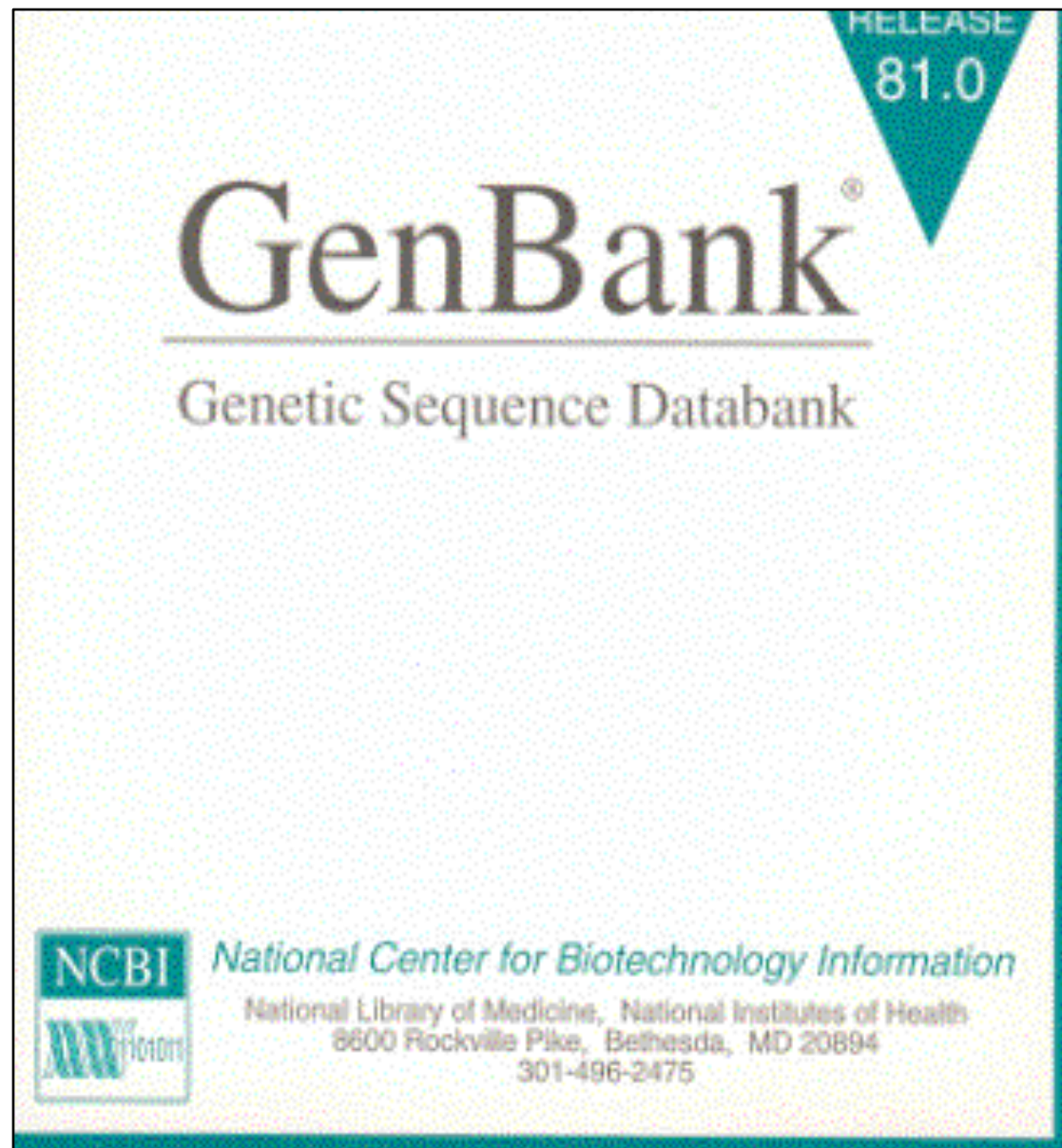
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```

## Flatfile format for INDSC:

- Header
- Feature Table: details about the sequence
- Sequence





An explosion of sequence data

<ftp://ftp.ncbi.nih.gov/genbank/>

Release 212.0	February 15, 2015
190,250,235	Reported Sequences
1,399,865,495,608	Total Bases

- full release every two months
- incremental updates daily
- available only via ftp

# Archival data

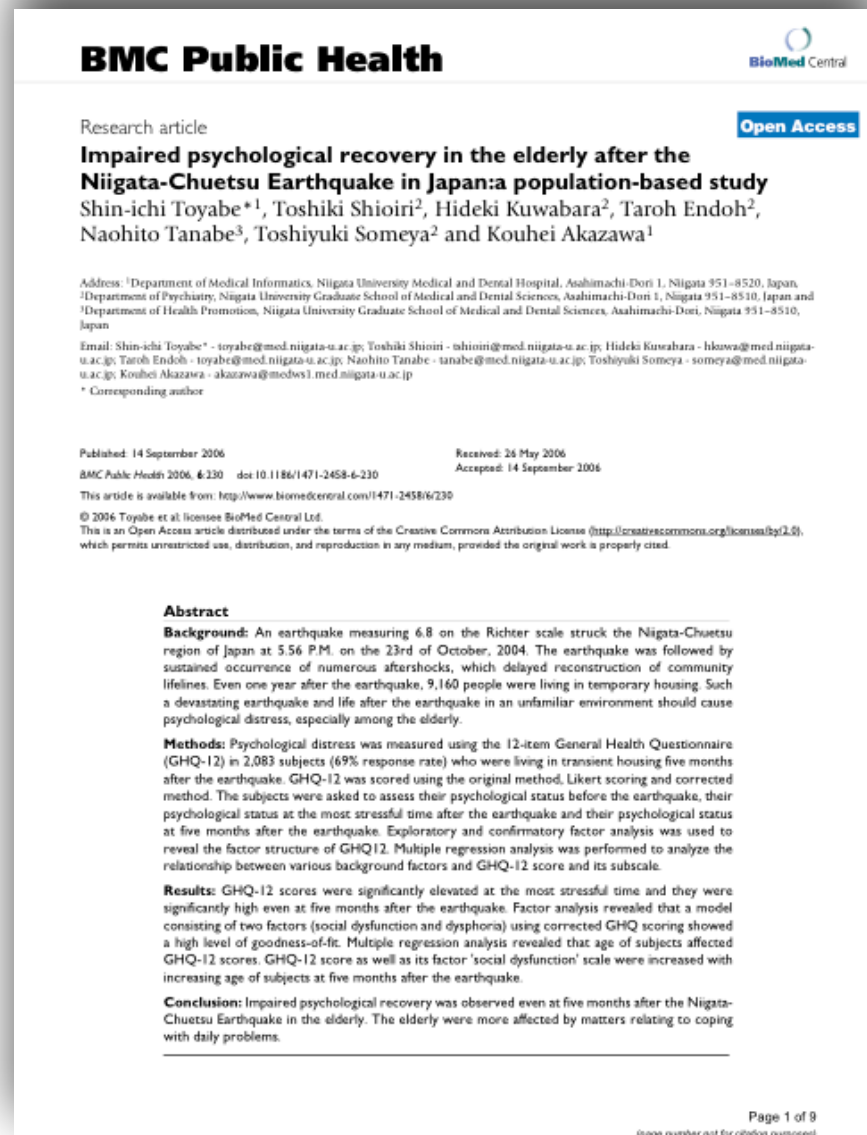
- ▶ repository of information
- ▶ redundant; might have many sequence records for the same gene, each from a different lab
- ▶ submitters maintain editorial control over their records:
- ▶ what goes in is what comes out
- ▶ no controlled vocabulary
- ▶ variation in annotation of biological features

# Curated data

- ▶ non-redundant; one record for each gene, or each splice variant
- ▶ each record is intended to present an encapsulation of the current understanding of a gene or protein
- ▶ records contain value-added information that have been added by an expert(s)

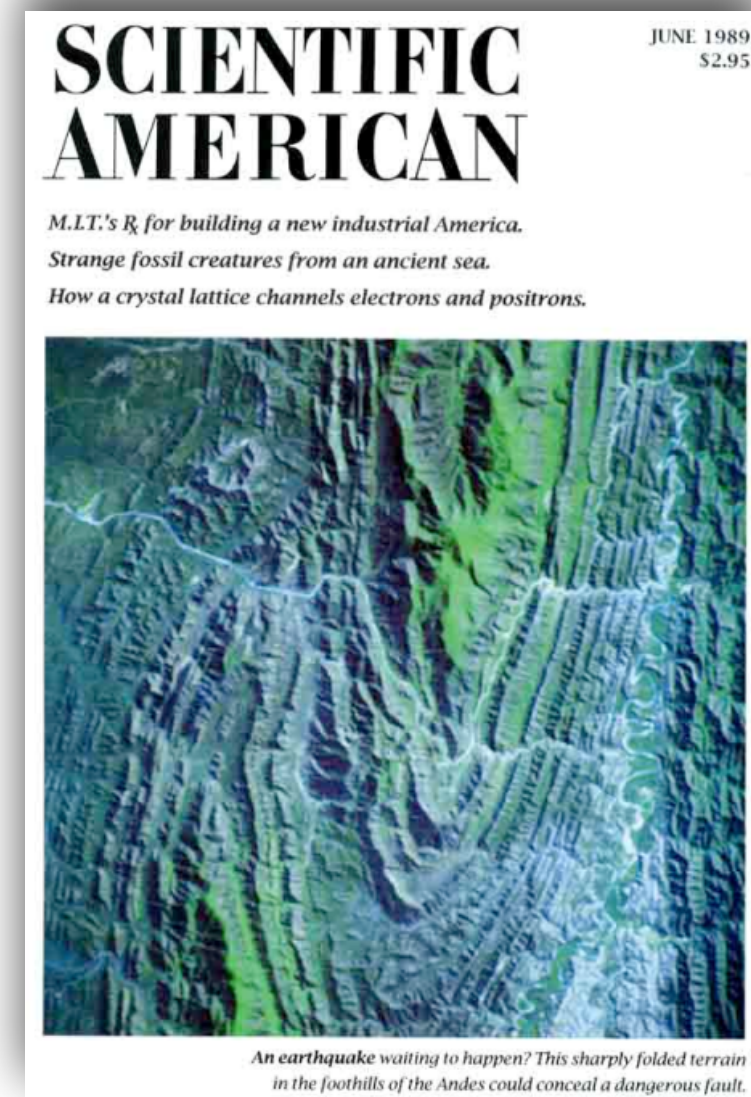


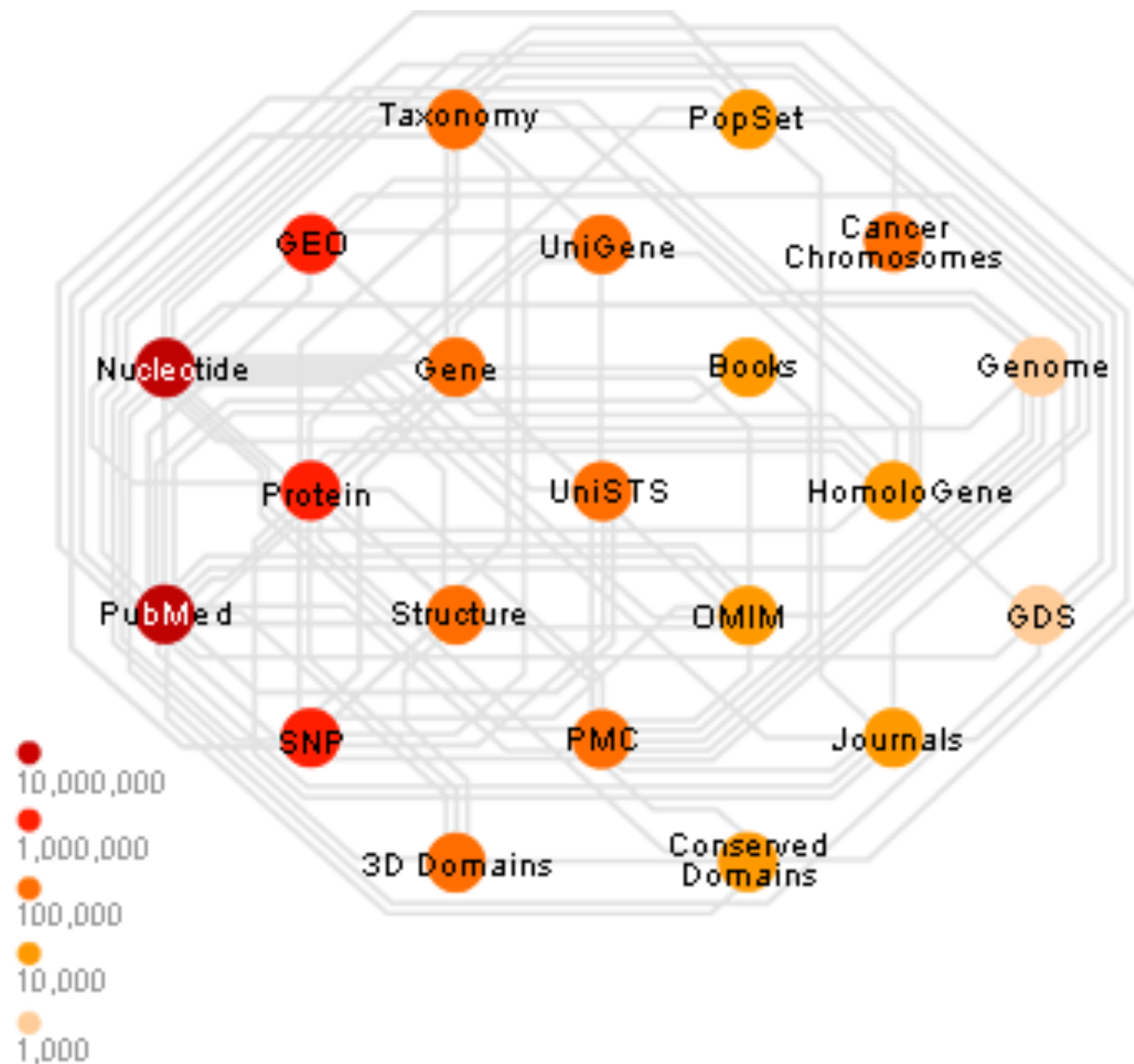
# Archival data



VS

# Curated data





More than just sequence data

# Genome assemblies

The genome sequence produced after chromosomes have been fragmented, sequenced and the resulting sequences have been put back together.

The **reference assembly** for a genome can be compiled from the DNA of one individual, a collection of individuals, a breed or a strain.





# Genome Reference Consortium

- ▶ “...working to create assemblies that better represent diversity and provide more robust substrates for genome analysis.”
  - ▶ novel assembly algorithm
  - ▶ correcting assembly errors (fix patches)
  - ▶ addition of new alternate loci (patches)
  - ▶ filling in gaps



GRCh37 or hg19?

# GRCh37 or hg19?

- ▶ Ensembl/NCBI versus UCSC
- ▶ chromosomal coordinates are the same
- ▶ contig sequences are the same, but different naming convention (i.e. 'chr1' versus '1')
- ▶ one-based coordinate system versus a zero-based coordinate system



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← Your awful, bigoted opinions are encoded  
in your genes

### Human species advised to move to GRCh37

Posted on [April 15, 2015](#) by [jovialscientist](#)

BOSTON. The entire human species has been advised to convert their genome to GRCh37 by the [GATK Best Practices team](#) at the Broad Institute, *The ScienceWeb* has learned.

GRCh37 is the *previous* version of the human genome reference. Last year, a rogue team of militant terrorist bioinformaticians within the Genome Reference Consortium released GRCh38, a hellish combination of core chromosomes, patches, unplaced contigs and alternate loci. In one fell swoop they broke every single bioinformatics pipeline ever written.

"Enough is enough" said Geraldine Van Damme, former martial arts expert and now head of the GATK team. "We took one look at GRCh38 and though 'that's it, we're sticking to GRCh37 and never moving'. We're therefore recommending that every human on the planet converts their genome to GRCh37. They should use CRISPR or something. It's going to make our lives a lot easier" she finished.

However, not everyone agrees. Deanna Cathedral, formerly Head of Anything Useful at the National Church of Biology Idiots (NCBI) said: "This reminds of the early days of the human genome project, when Frankie Collins suggested we try and genetically modify everyone to be haploid. It's just not realistic" she concluded.

#### Recent Posts

- [Human species advised to move to GRCh37](#)
- [Your awful, bigoted opinions are encoded in your genes](#)
- [Only three gel images ever made, admit scientists](#)
- [Bacteria will pay you to sequence them by 2016, analysis reveals](#)
- [SGM held at Birmingham to allow scientists to collect filthy new diseases](#)

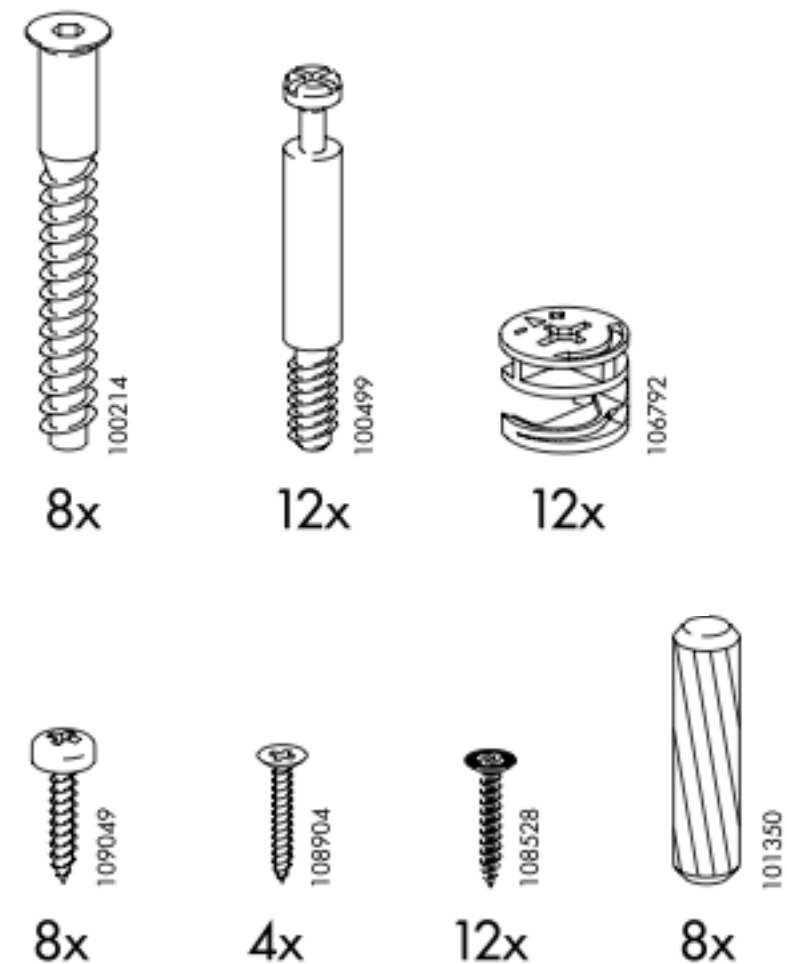
#### Meta

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- [Comments RSS](#)
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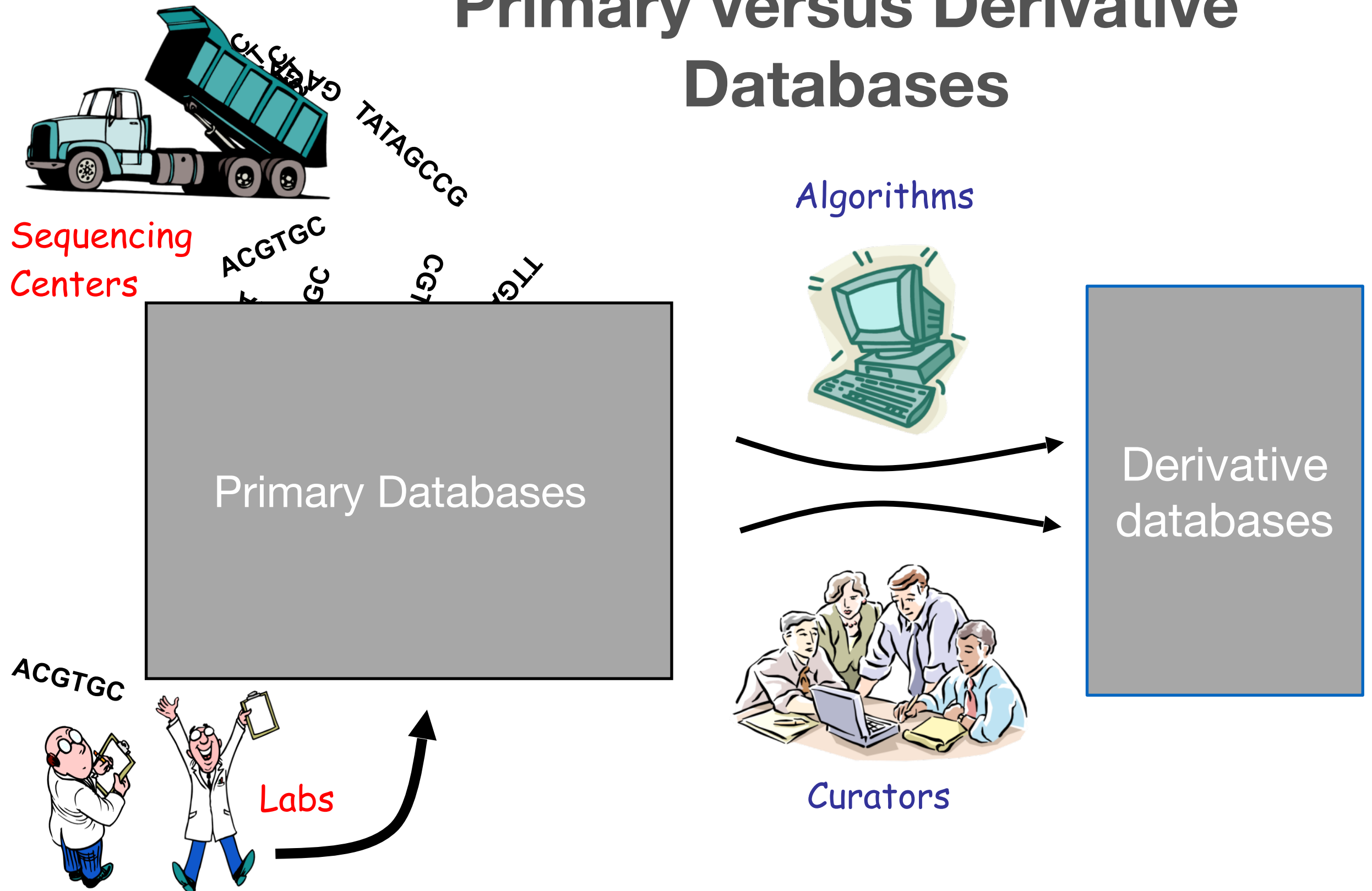
# Gene Builds

(not to be confused with *genome* builds)

- ▶ A set of annotations for the assembled genome
- ▶ Database specific
- ▶ Predicted genes based on varying levels of evidence



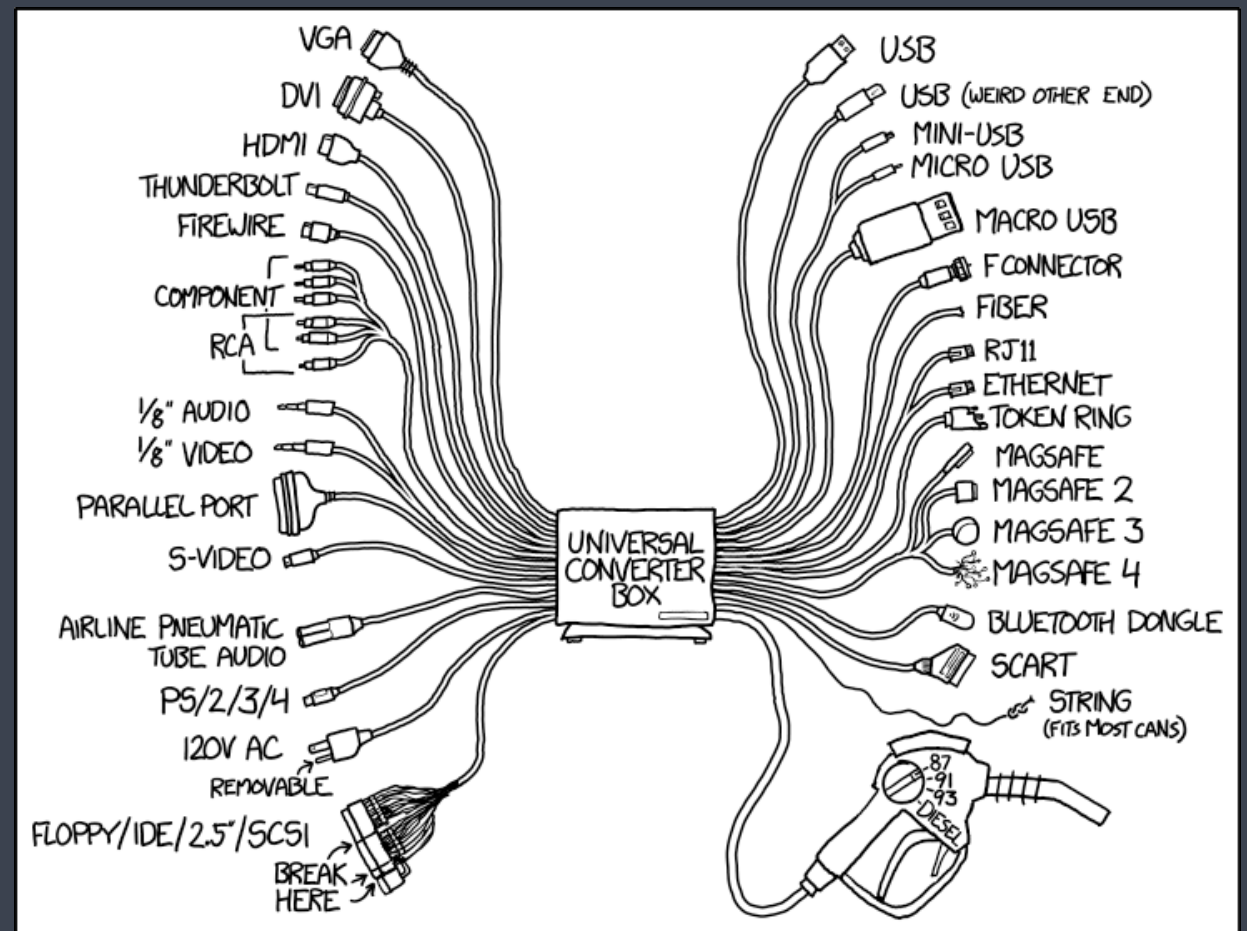
# Primary versus Derivative Databases





# ID conversions: Biomart

- ▶ a search engine (part of Ensembl) that can map terms across multiple domains and output them into table format
- ▶ No programming required
  1. Choose database
  2. Apply filters (which genes do we want to look at)
  3. Choose attributes (determine output columns)



# Genome Browsers

- ▶ a graphical interface for display of information from a biological database for genomic data
  - ▶ General use (UCSC, IGV)
  - ▶ Human-specific; Other vertebrate genomes (Ensembl, Entrez)
  - ▶ Non-vertebrate genome (FlyBase, WormBase, TAIR)



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