Sequence Tracking

Understanding your sequence context

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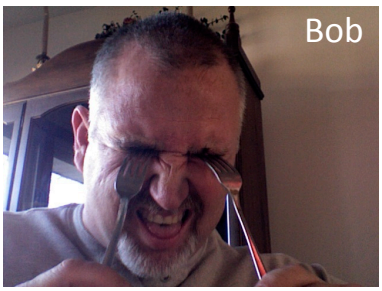
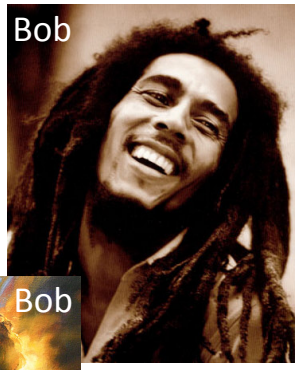
Personalis, Inc

Personalis

What's in a name?



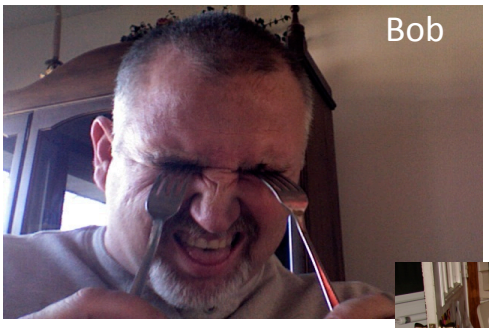


What's in a name?

123-45-6789

*http://howmanyofme.com

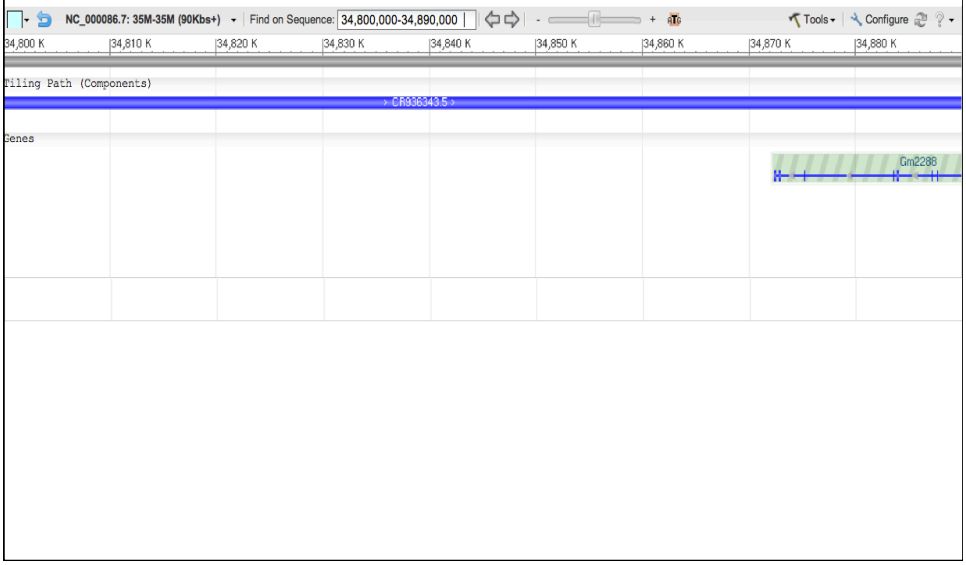
What's in a name?



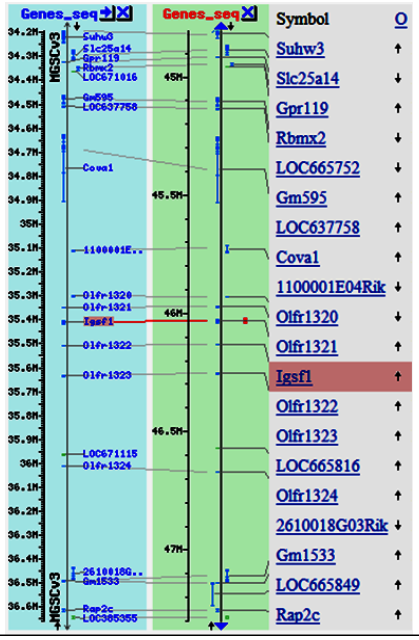
Need more than unique identifier
track updates/improvements

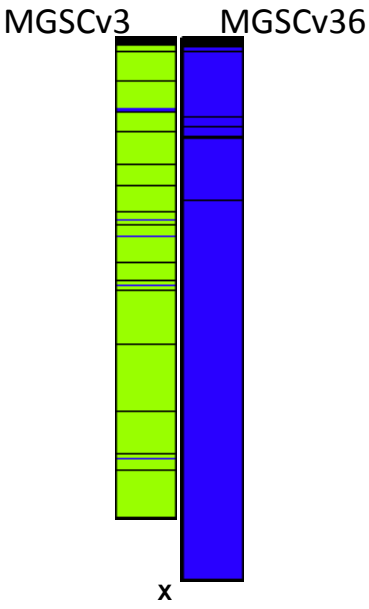
chr1
Chr1
1
Chrom1

Mouse chrX: 34,800,000-34,890,000

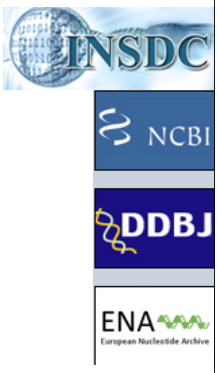


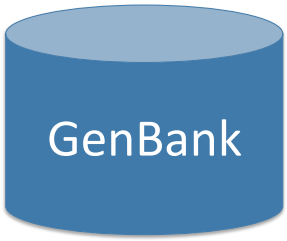
Mouse chrX: 35,000,000-36,000000





Data Archives





- Data in a common format
- Data in a single location (and mirrored)
- Most quality checked prior to deposition
- Robust data tracking mechanism (accession.version)
- Data owned by submitter

Data tracking

ABC14-1065514J1

	Date	Phase	Gaps	Length
FP565796.1	21-Oct-2009	1	1	
FP565796.2	14-Oct-2010	1	0	
FP565796.3	07-Nov-2010	3	0	

Data Archives

Initial versions of human and mouse reference assemblies not in INSDC!!*

First human version in INSDC: GRCh37
First mouse version in INSDC: NCBI36

* But were tracked by RefSeq

Data Archives

INSDC archives track INDIVIDUAL sequences

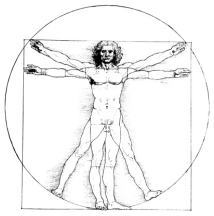
Homo sapiens chromosome 9 genomic contig, **GRCh37** reference primary assembly
3,818,133 bp linear DNA
Accession: GL000090.1 GI: 224183256
GenBank FASTA Graphics

Homo sapiens chromosome 9 genomic contig, **GRCh37** reference primary assembly
62,237,592 bp linear DNA
Accession: GL000089.1 GI: 224183255
GenBank FASTA Graphics

Homo sapiens chromosome 9 genomic contig, **GRCh37** reference primary assembly
178,933 bp linear DNA
Accession: GL000088.1 GI: 224183254
GenBank FASTA Graphics

An assembly is a COLLECTION of sequences

More naming issues



GRCh37

hg19

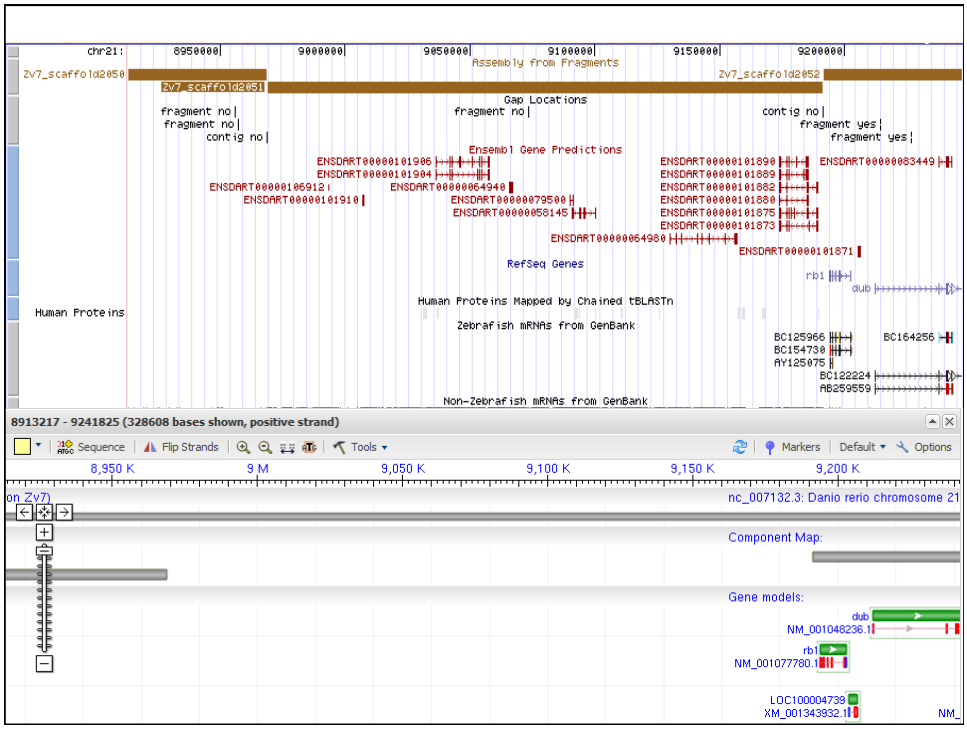
Zv7

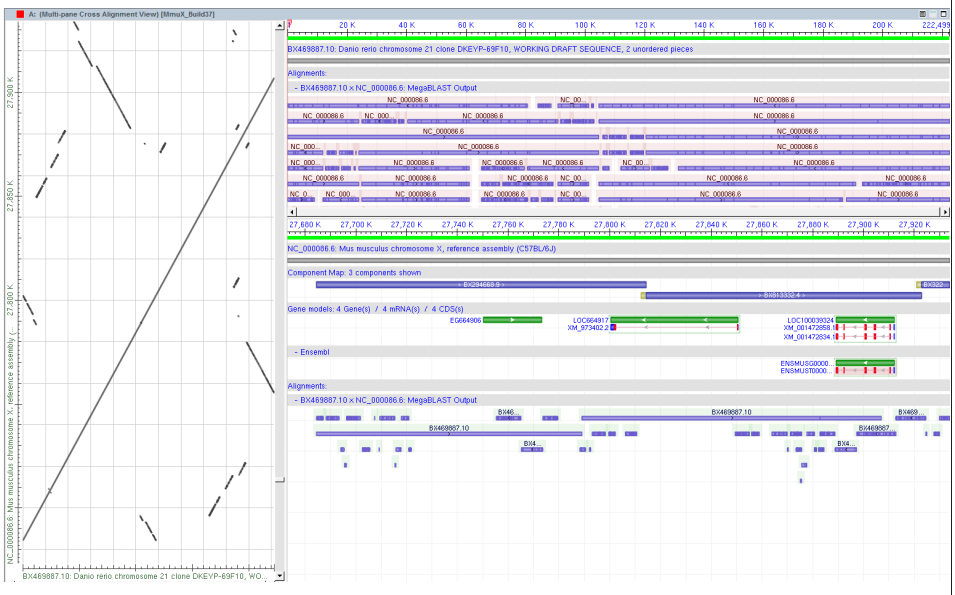
danRer5

MGSCv37

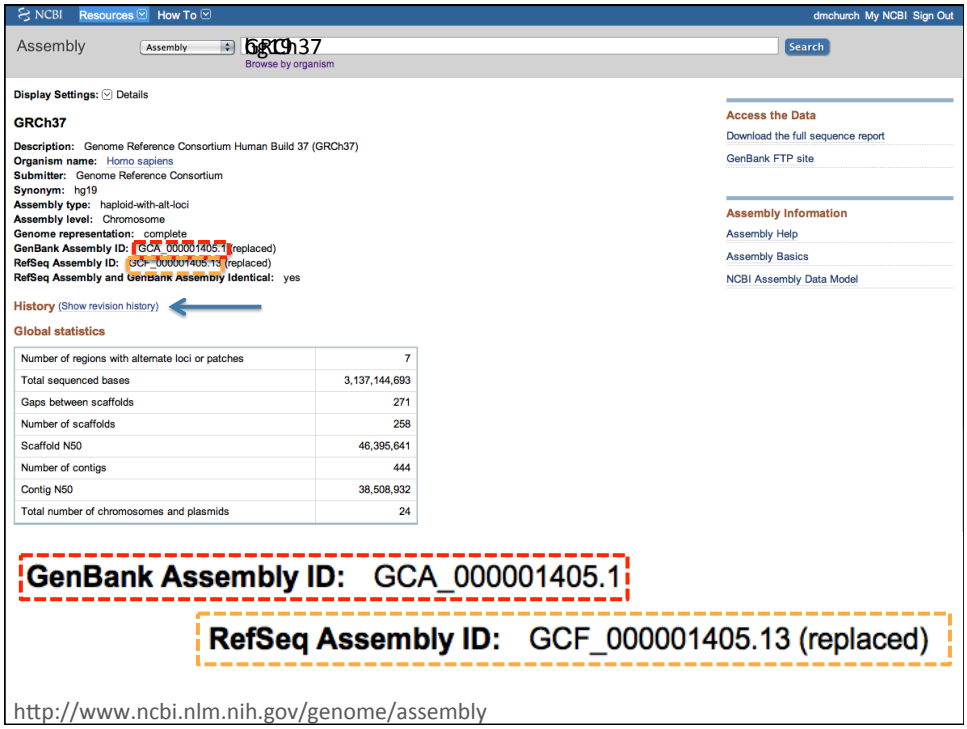
mm8

NCBIM37





Zv7 chr21:8,913,216-9,246,964 vs MGSCv36 chrX

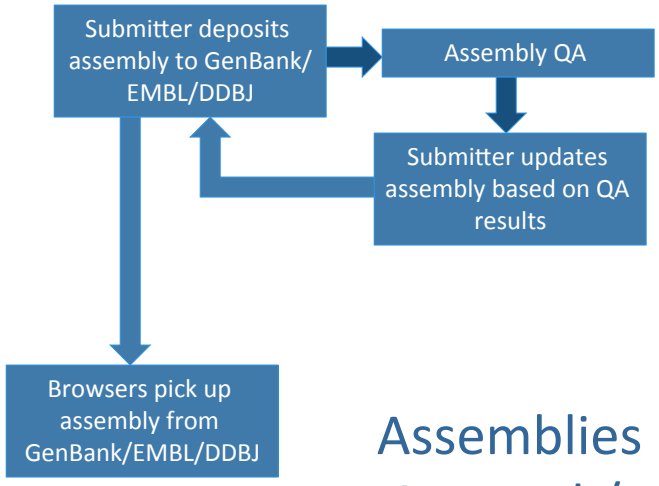


http://www.ncbi.nlm.nih.gov/genome/assembly

History (Hide revision history)

GenBank Assembly Accession		RefSeq Assembly Accession	Assembly Name	Assembly Level	Status
GCA_000001405.8	n/a	n/a	**GRCh37.p7**	**Chromosome**	**Latest GenBank**
GCA_000001405.7	n/a	n/a	GRCh37.p6	Chromosome	Replaced GenBank
GCA_000001405.6	=	GCF_000001405.17	GRCh37.p5	Chromosome	Latest RefSeq, Replaced GenBank
GCA_000001405.5	n/a	n/a	GRCh37.p4	Chromosome	Replaced GenBank
GCA_000001405.4	n/a	n/a	GRCh37.p3	Chromosome	Replaced GenBank
GCA_000001405.3	=	GCF_000001405.14	GRCh37.p2	Chromosome	Replaced GenBank, Replaced RefSeq
GCA_000001405.2	n/a	n/a	GRCh37.p1	Chromosome	Replaced GenBank
GCA_000001405.1	=	GCF_000001405.13	GRCh37	Chromosome	Replaced GenBank, Replaced RefSeq
n/a	n/a	GCF_000001405.12	NCBI36	Chromosome	Replaced RefSeq
n/a	n/a	GCF_000001405.11	NCBI35	Chromosome	Replaced RefSeq
n/a	n/a	GCF_000001405.10	NCBI34	Chromosome	Replaced RefSeq
n/a	n/a	GCF_000001405.9	NCBI34	Chromosome	Replaced RefSeq

Genome Browser Agreement



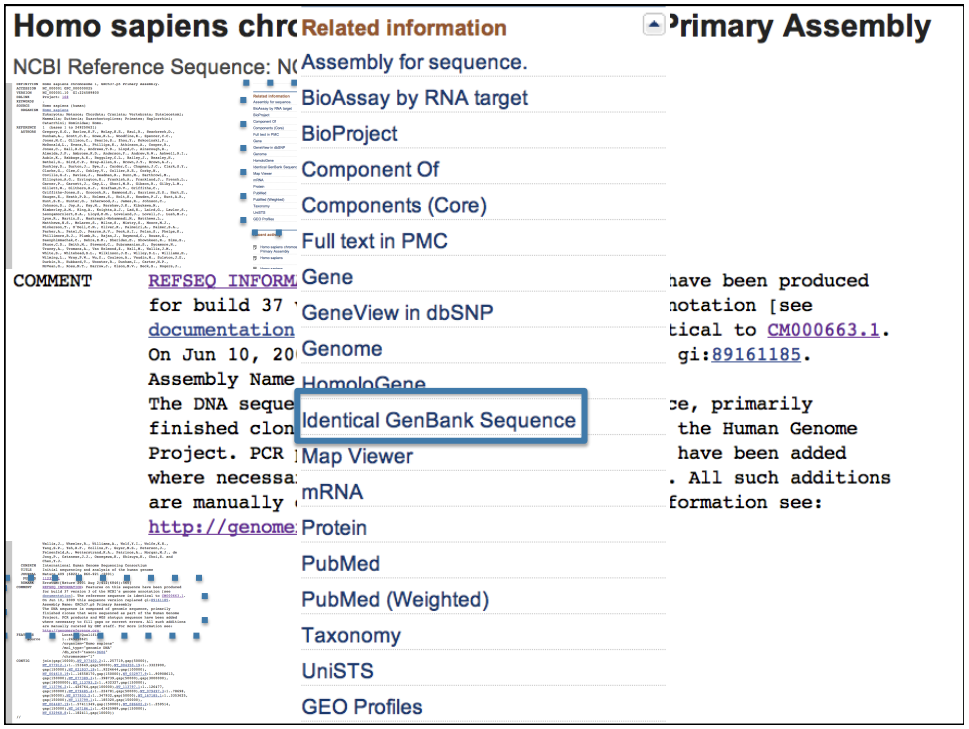
Assemblies must be in GenBank/EMBL/DDBJ

e!Ensembl NCBI UCSC Genome Bioinformatics

GenBank	vs	RefSeq
Submitter Owned		RefSeq Owned
Redundancy		Non-Redundant
Updated rarely		Curated
INSDC		Not INSDC

BRCA1

83 genomic records	3 genomic records
31 mRNA records	5 mRNA records
27 protein records	1 RNA record
	5 protein records



RefSeq for Assemblies

Typical assembly edits

Addition of non-nuclear (e.g. MT) assembly units

Removal of contamination

Drop unlocalized/unplaced scaffolds
Mask contamination that is placed on chromosome (while preserving coordinate space)

Human assemblies in assembly database

Organism	Name	Submitter	Date	Genome representation	Assembly level	Version status	Representative status
Homo sapiens	CHM1_1.1	Washington University School of Medicine	2013/06/14	full	Chromosome	latest	na
Homo sapiens	YH_2.0	Beijing Genomics Institute	2013/06/10	full	Scaffold	latest	na
Homo sapiens	WGSA	Celera Genomics	2004/02/25	full	Chromosome	latest	na
Homo sapiens	CSA	Celera Genomics	2004/02/25	full	Chromosome	latest	na
Homo sapiens	HuRefPrime	J. Craig Venter Institute	2008/09/24	full	Chromosome	latest	na
Homo sapiens	HsapALLPATHS1	Broad Institute	2011/01/06	full	Scaffold	latest	na
Homo sapiens	Watson-partial	Baylor College of Medicine	2008/04/17	partial	Contig	latest	na
Homo sapiens	BGIAF	Beijing Genomics Institute	2010/06/21	full	Scaffold	latest	na
Homo sapiens	GRCh38 UCSC Name: hg38	Genome Reference Consortium	2013/12/17	full	Chromosome	latest	representative-genome
Homo sapiens	RP11_1.0_unmatched_regions	Roche	2013/07/31	partial	Scaffold	latest	na
Homo sapiens	CRA_TCAGchr7v2	The Centre for Applied Genomics	2004/09/01	partial	Chromosome	latest	na
Homo sapiens	HuRef	J. Craig Venter Institute	2007/09/24	full	Chromosome	latest	na

http://www.ncbi.nlm.nih.gov/assembly/organism/9606/

Take home messages

- Assemblies can (and do) update!
- Know what assembly your are working on
  - Track by accession.version, not just name
- Data in INSDC databases are mirrored
- RefSeq is NCBI specific