Ensembl

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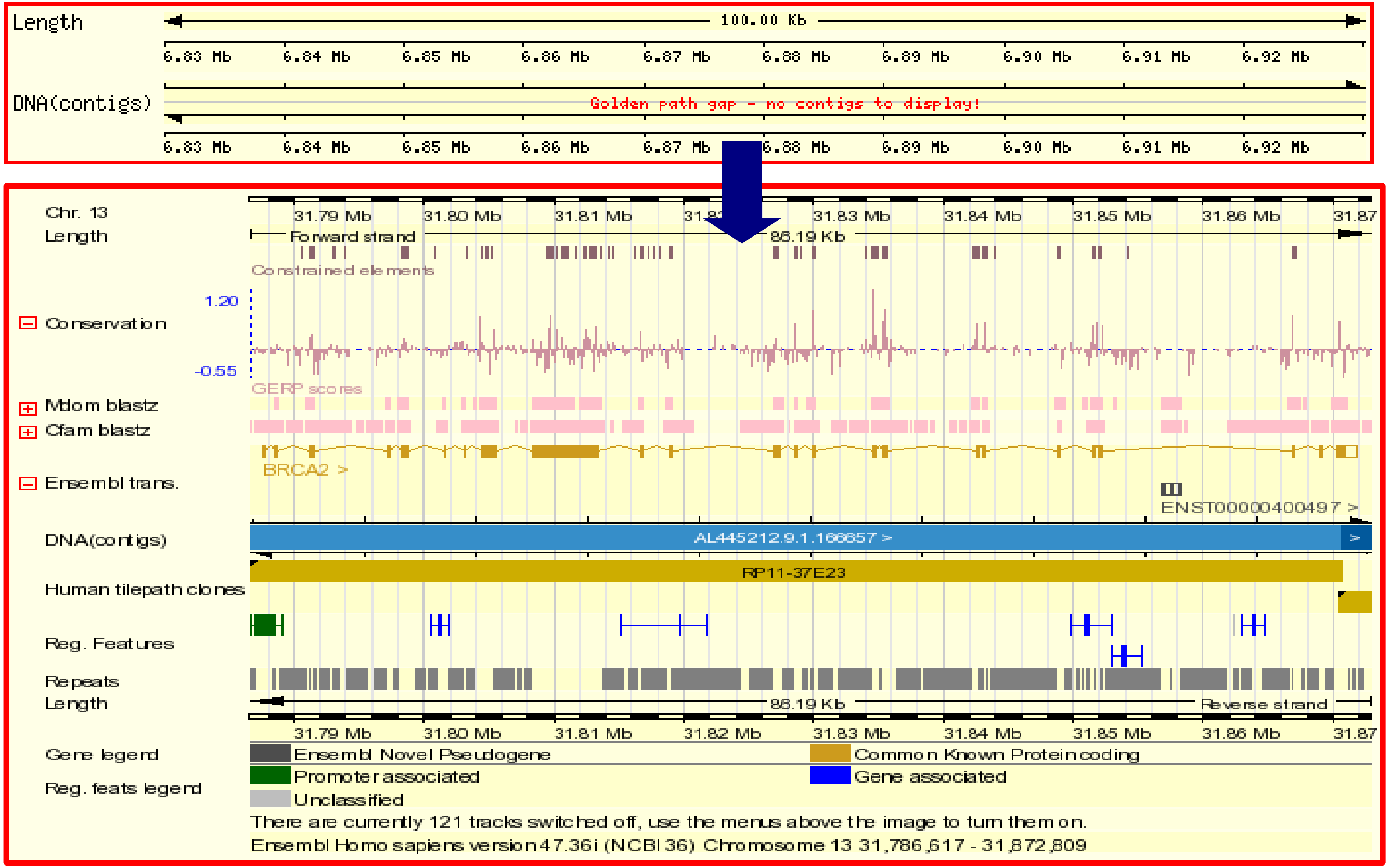
EMBL-EBI

wellcome trust
sanger
institute

Ensembl mission

To enable genomic science by providing high-quality, integrated annotation on vertebrate genomes within a consistent and accessible infrastructure.



There are currently 121 tracks switched off, use the menus above the image to turn them on.

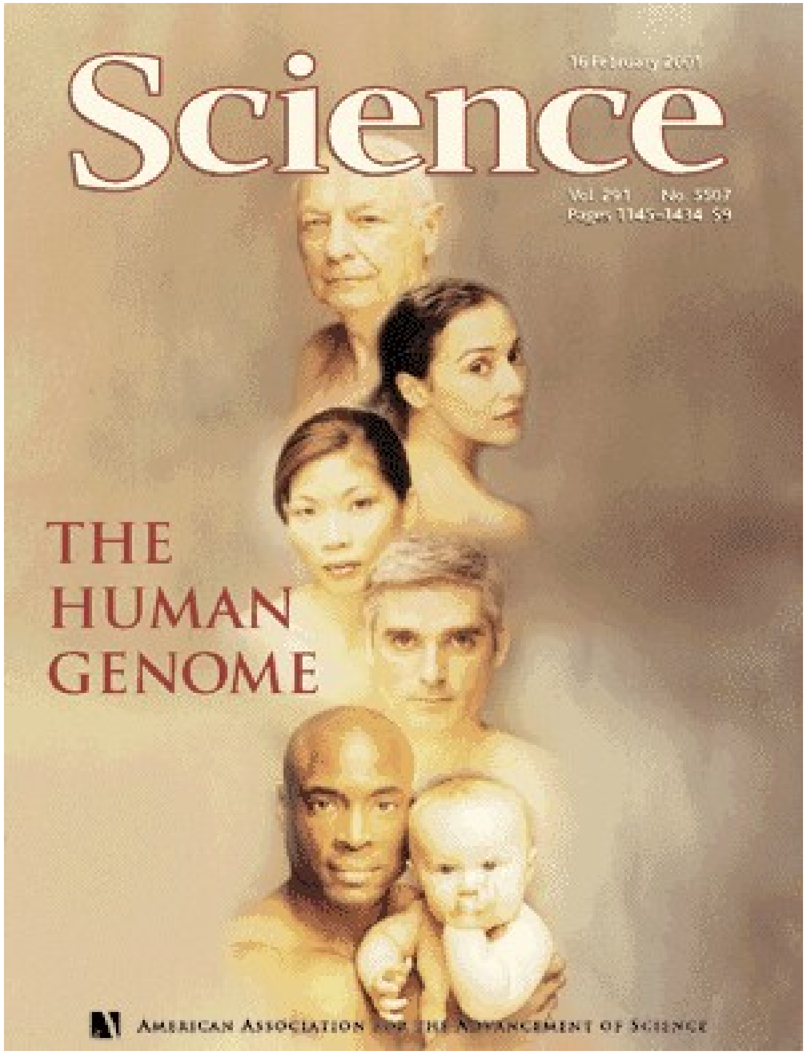
Ensembl Homo sapiens version 47.36i (NCBI 36) Chromosome 13 31,786,617 - 31,872,809

Ensembl Concept

- Collaborative project of the European Bioinformatics Institute (EBI) and the Wellcome Trust Sanger Institute
- Provides annotation and analysis of chordate genomes
- Open by design
  - Code is BSD, not GNU
  - All data is freely available
- Continuously developed and comprehensively updated 5 times a year
- Diverse skills across the project
- Technology adopted and used by many other projects

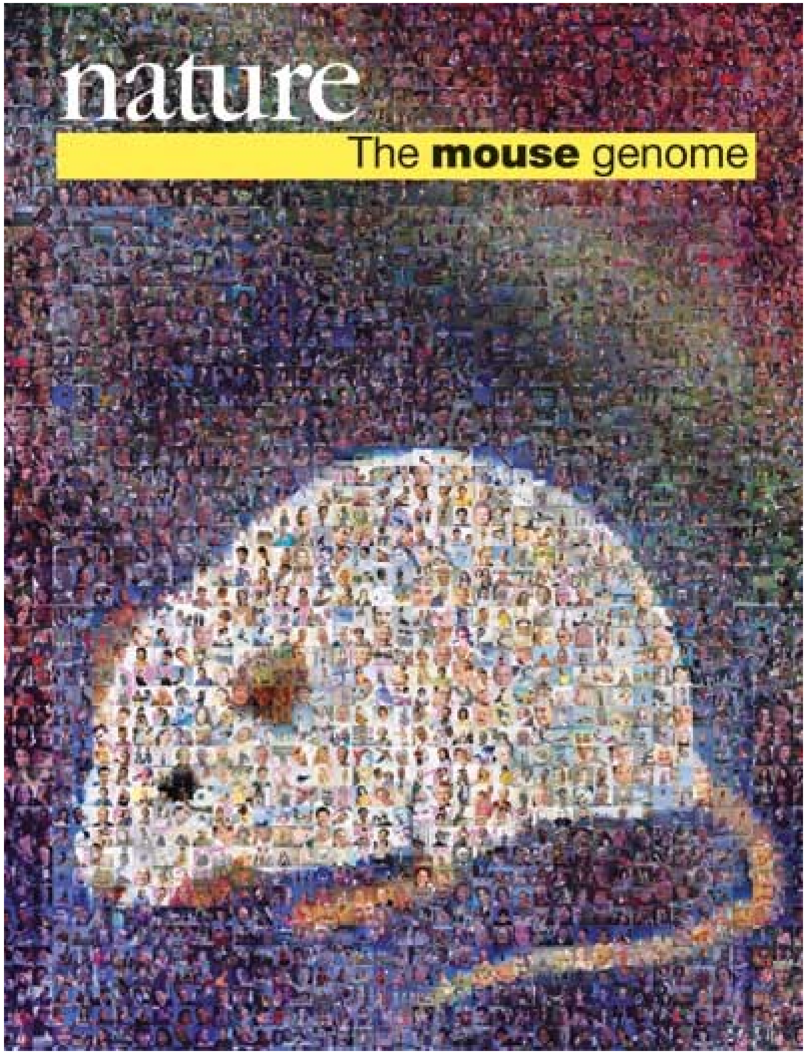
Human genome (Feb 2001)

Nature, 15th Feb 2001



Science, 15th Feb 2001

Mouse genome (Dec 2002)



- 2nd mammal genome
- model organism in lab
- 14% smaller than human
- 40% can be aligned to human
- 5% under purifying selection
- 0.5 substitutions per site, twice as many in the mouse lineage

Nature 420, 520-562

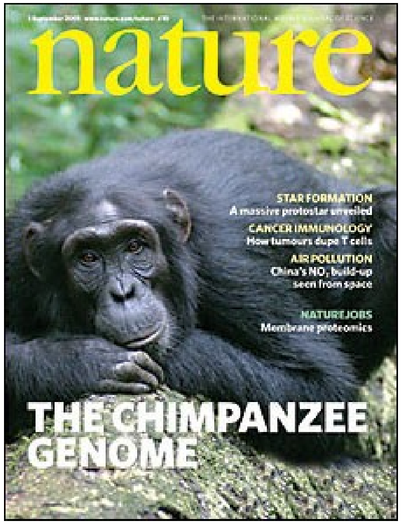
Rat genome (Apr 2004)

- 3rd mammal, 2nd rodent
- Similar number of genes in all 3 species
- 40% eutherian specific seq.
- 30% rodent specific seq., mostly repeats
- At least half of unaligned seq. is rat-specific repeats

Nature 428, 493-521

More and more genomes

2004

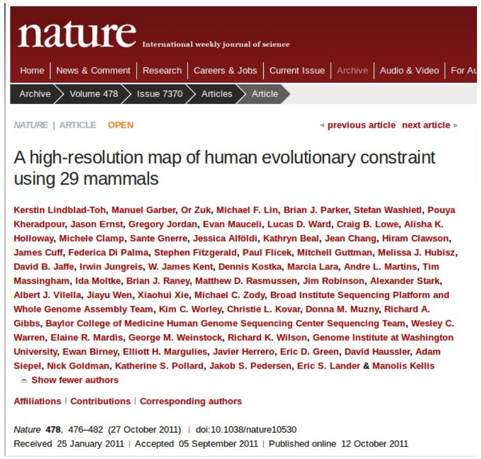


2005

2007

nature
CARBON DIOXIDE CAPTURE
Cagey ZIFs for emissions control
'FLIPPING' ENZYMES
Action mechanism of HIV's Achilles' heel
SCIENCE AND MUSIC
First in a new series
THE PLATYPUS GENOME
Sequence analysis reveals clues to early mammalian evolution
NATUREJOBS
Up Canada way

2008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A high-resolution map of human evolutionary constraint using 29 mammals

Kerstin Lindblad-Toh, Manuel Garber, Or Zuk, Michael F. Lin, Brian J. Parker, Stefan Washietl, Pouya Kheradpour, Jason Ernst, Gregory Jordan, Evan Mauceli, Lucas D. Ward, Craig B. Lowe, Alisha K. Holloway, Michele Clamp, Sante Gnerre, Jessica Alföldi, Kathryn Beal, Jean Chang, Hiram Clawson, James Cuff, Federica Di Palma, Stephen Fitzgerald, Paul Flicek, Mitchell Guttman, Melissa J. Hubisz, David B. Jaffe, Irwin Jungreis, W. James Kent, Dennis Kostka, Marcia Lara, Andre L. Martins, Tim Massingham, Ida Moltke, Brian J. Raney, Matthew D. Rasmussen, Jim Robinson, Alexander Stark, Albert J. Vilella, Jiayu Wen, Xiaohui Xie, Michael C. Zody, Broad Institute Sequencing Platform and Whole Genome Assembly Team, Kim C. Worley, Christie L. Kovar, Donna M. Muzny, Richard A. Gibbs, Baylor College of Medicine Human Genome Sequencing Center Sequencing Team, Wesley C. Warren, Elaine R. Mardis, George M. Weinstock, Richard K. Wilson, Genome Institute at Washington University, Ewan Birney, Elliott H. Margulies, Javier Herrero, Eric D. Green, David Haussler, Adam Siepel, Nick Goldman, Katherine S. Pollard, Jakob S. Pedersen, Eric S. Lander & Manolis Kellis

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Affiliations | Contributions | Corresponding authors

Nature **478**, 476–482 (27 October 2011) | doi:10.1038/nature10530

Received 25 January 2011 | Accepted 05 September 2011 | Published online 12 October 2011

2011

More than 50 vertebrate genomes have been “fully” sequenced

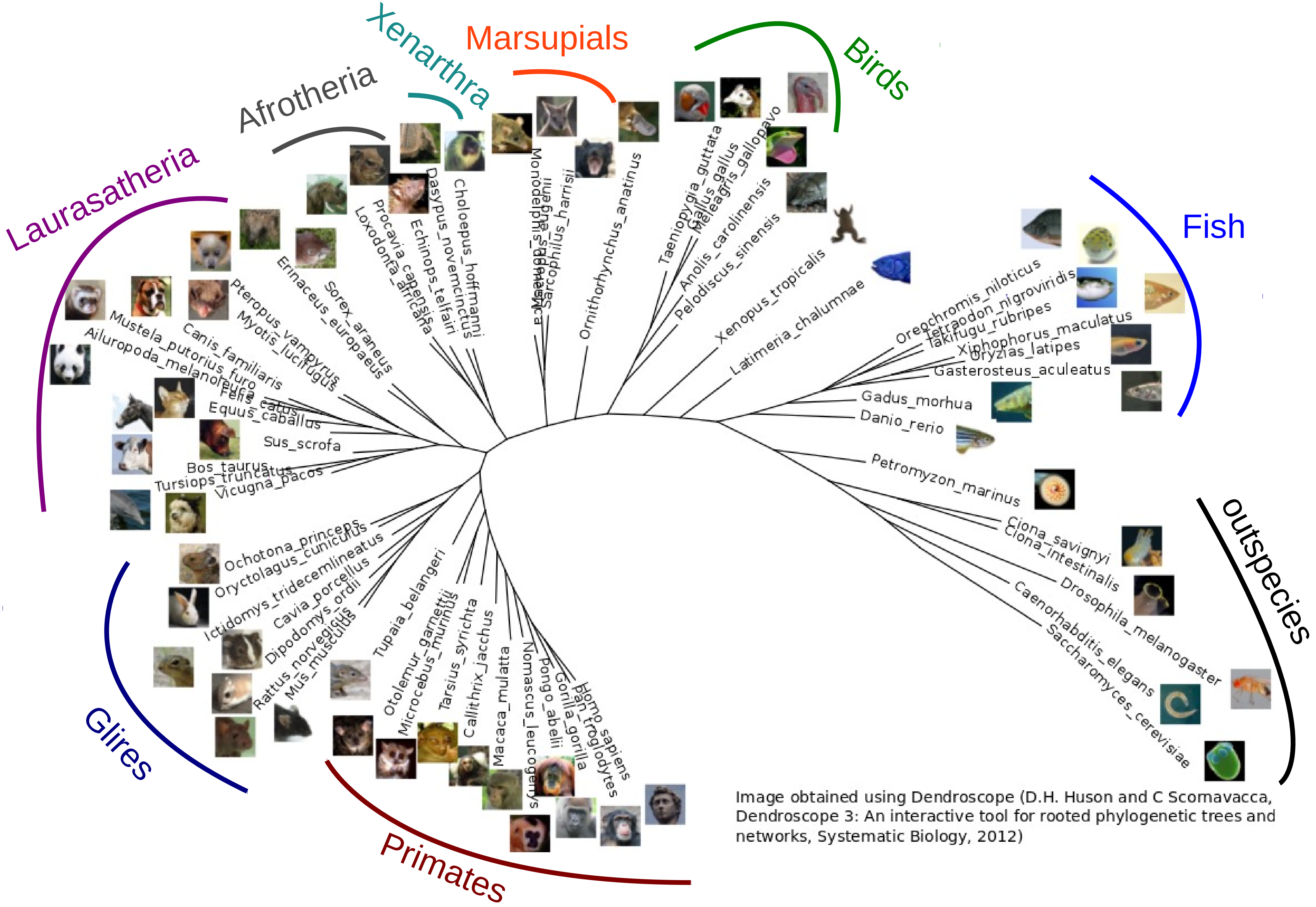


Image obtained using Dendroscope (D.H. Huson and C Scornavacca, Dendroscope 3: An interactive tool for rooted phylogenetic trees and networks, Systematic Biology, 2012)

1000 genomes pilot project (Oct 2010)

nature
A THOUSAND GENOMES
Pilot studies prepare the way for population-scale gene sequencing PAGES 1050 & 1061

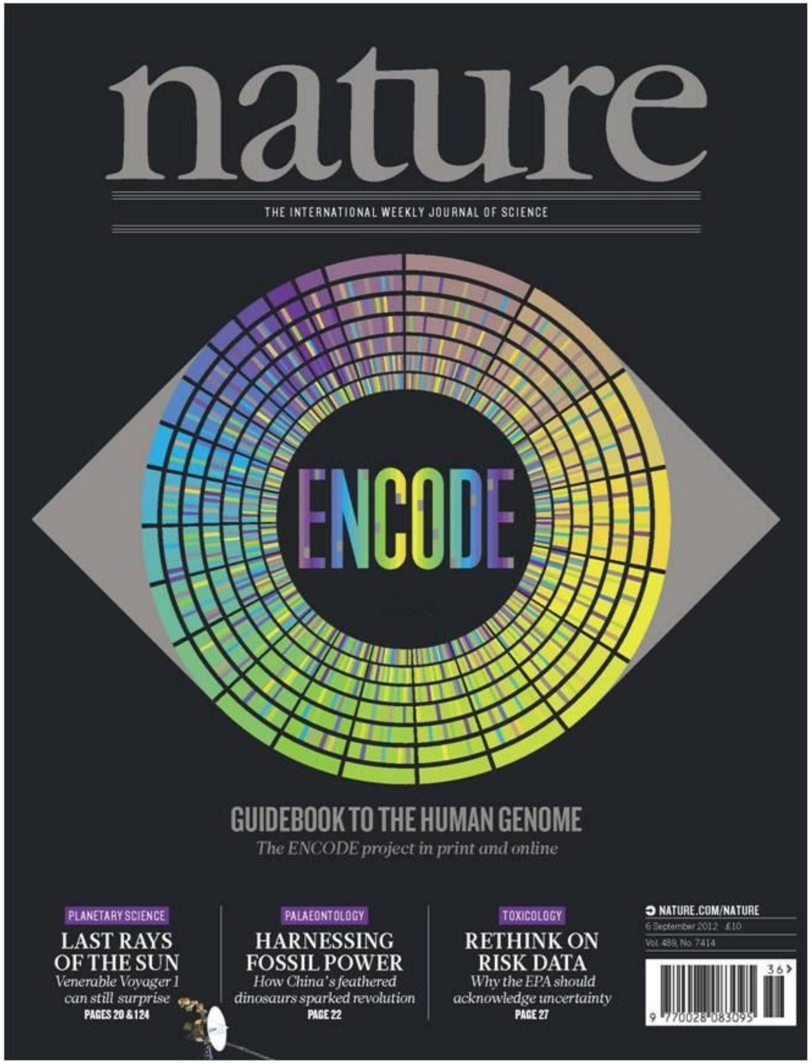
3 pilots:

- 179 ind. low-cov
- 2 trios high-cov
- 679 ind. exon only

Covers >95% of variants of any individual

Each individual carries 250-300 loss-of-function variants; 50-100 implicated in inherited disorders

ENCODE project: ENCyclopedia Of DNA Elements


nature
THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE
ENCODE
GUIDEBOOK TO THE HUMAN GENOME
The ENCODE project in print and online
PLANETARY SCIENCE
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Why the EPA should acknowledge uncertainty
PAGE 27
NATURE.COM/NATURE
6 September 2012 £10
Vol 489, No 7414

Nature 489, 57–74 (06 Sept 2012)

- 1640 data sets
- 147 different cell types
- 400+ authors

- Activity in 80% of the genome
- Evidence of negative selection (in aggregate) in primate-specific elements
- Could classify the genome into 7 different chromatin states
- Transcription ↔ Histone + TF

Ensembl: What do you get?

Genome Annotation

- Protein coding gene structure
  - Consistent with genome, predicted across all vertebrates
  - Manual annotations (human, mouse, zebrafish, MHC)
- RNA genes (including miRNA)
  - Consistent with genome, predicted in across mammals
- Additional identifiers per genes (Xref)
  - Affymetrix, EntrezGene, Uniprot…

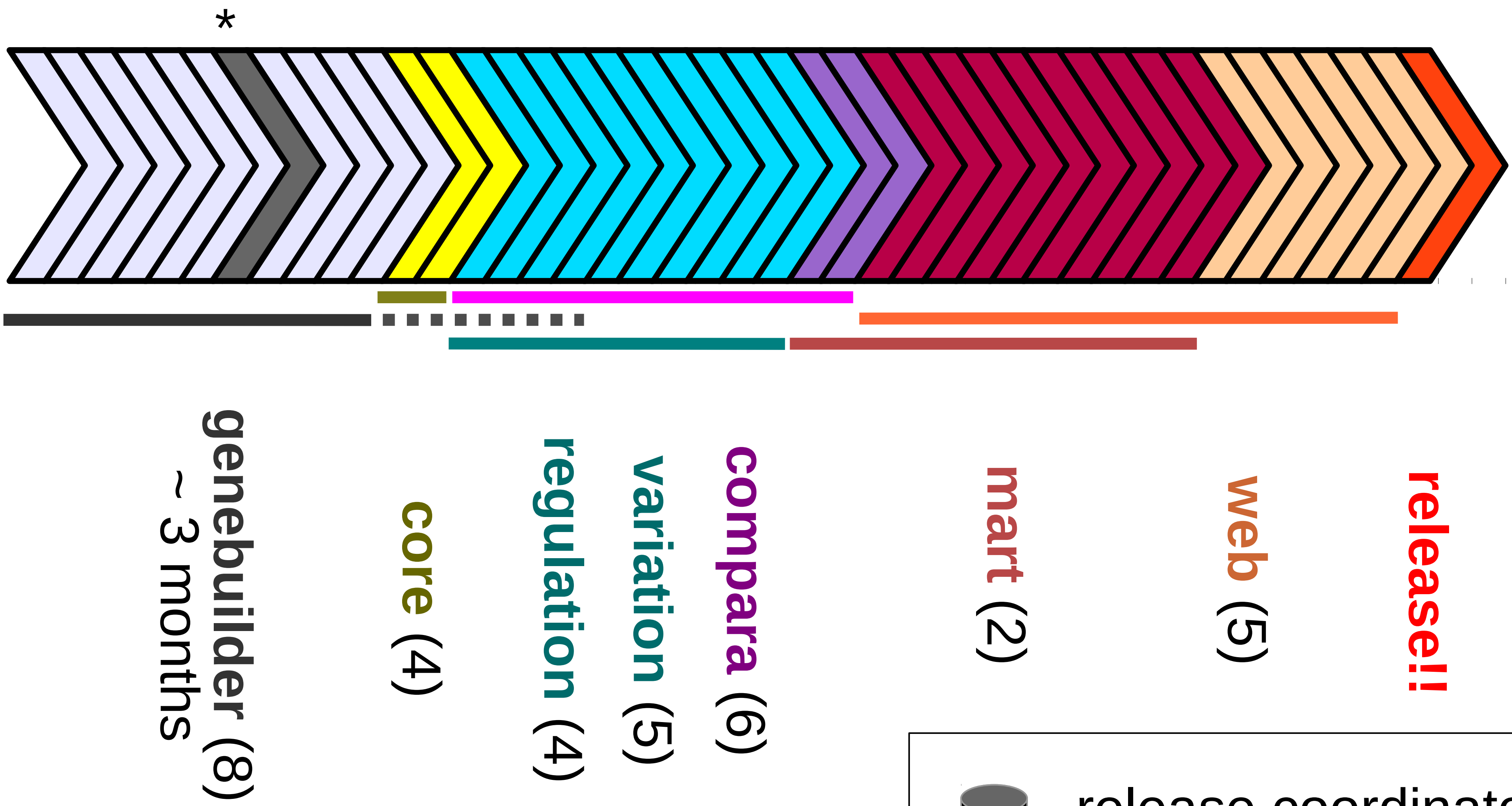
Variation, Comparative & Functional Genomics

- Genome alignments
  - Blastz, Blat, Pecan (multiple alignments), EPO
  - Homologues between genomes
  - Protein trees
- Variants (SNPs), CNVs, strains, genotypes
- ChIP-chip, ChIP-seq, segmentations

Infrastructure

- Website, Data mining tool, database and data dumps
- Portable, extendible, open source system with database, DAS, API, website, pipeline

Ensembl release cycle

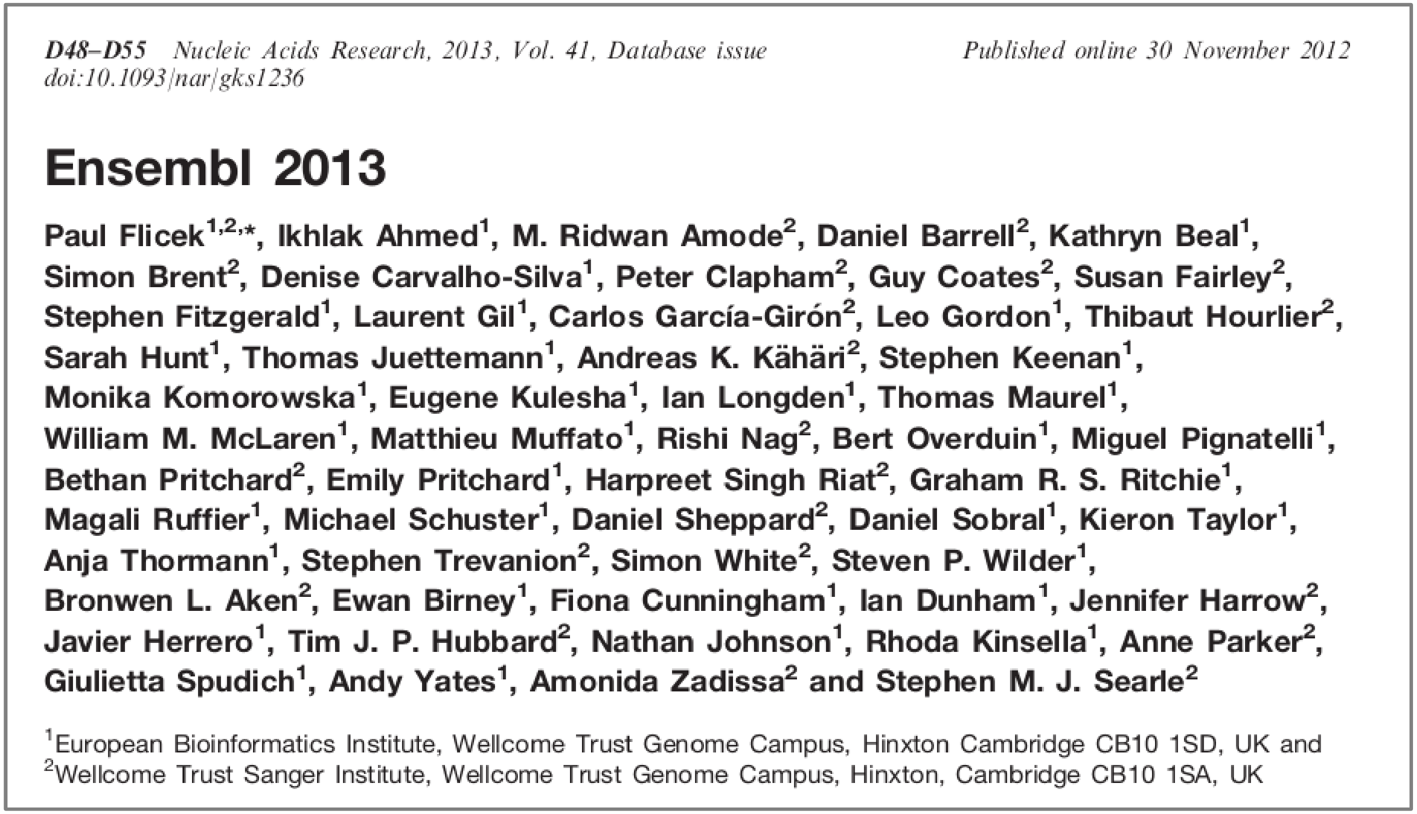


release coordinator
+ assistant

Ensembl groups

- **GeneBuilders**: sequence masking, gene building
- **Core**: database schema, stable id mapping
- **Compara**: protein homology, genomic sequence alignments
- **Variation**: SNPs, CNVs, personal/strain genomes
- **Regulation**: probe mapping, functional data, segmentation
- **Web**: web site, new views for new data
- **Outreach**: help, workshops, tutorials
- **Production**: BioMart, coordination
- More people: Research, e! genomes, Zebrafish, Systems...

Acknowledgements



D48–D55 *Nucleic Acids Research, 2013, Vol. 41, Database issue* *Published online 30 November 2012*
doi:10.1093/nar/gks1236

Ensembl 2013

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EMBL

National Human Genome Research Institute

BBSRC bioscience for the future

European Commission
Framework Programme 7

SEVENTH FRAMEWORK PROGRAMME

GEN2PHEN

BLUEPRINT epigenome

Quantomics
From Sequence to Consequence :
Tools for the Exploitation of Livestock Genomes

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