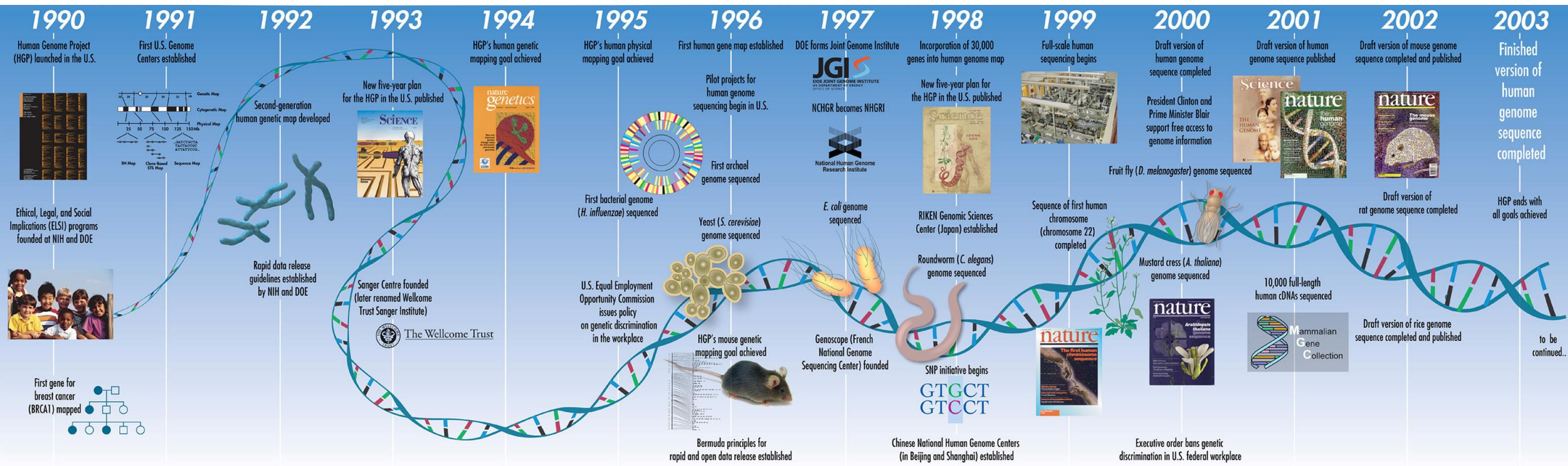




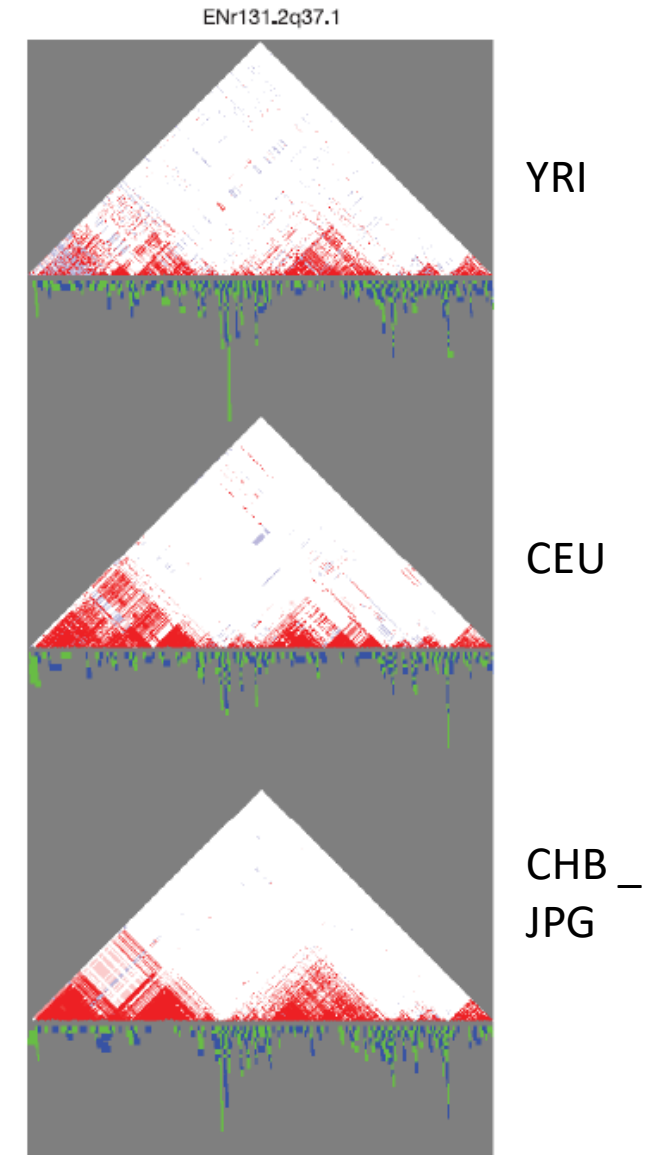
Introduction to common variation (II)

Lucía Colodro Conde and Katrina Grasby



Projects to create reference panels have provided information on:

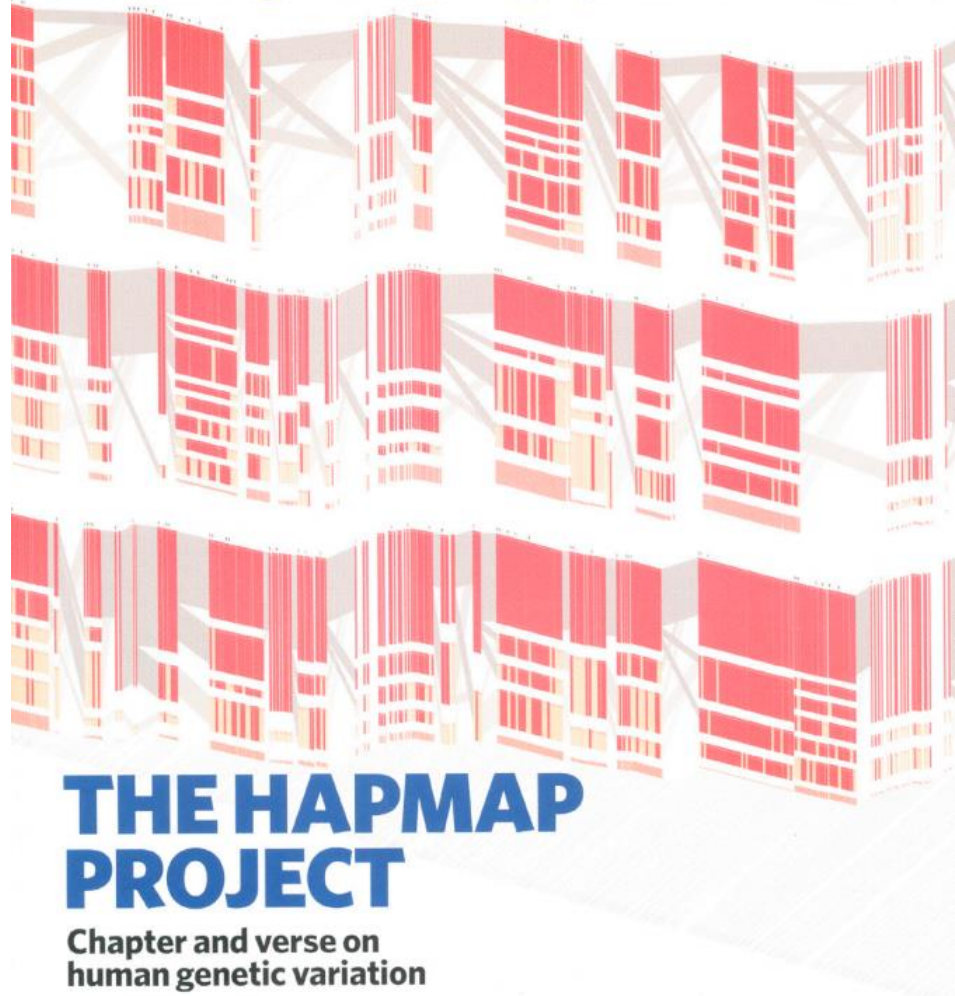
- Patterns of human common genetic variation
- Linkage disequilibrium (LD) and allele frequencies differences in populations
- Tag SNPs for the design of SNP arrays to facilitate imputation and GWAS



27 October 2005 | www.nature.com/nature

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

nature



HapMap (haplotype map) Project

270 individuals:

30 parent-offspring trios of the Yoruba from Ibadan, Nigeria (YRI)

30 trios of Utah residents with European ancestry (CEU)

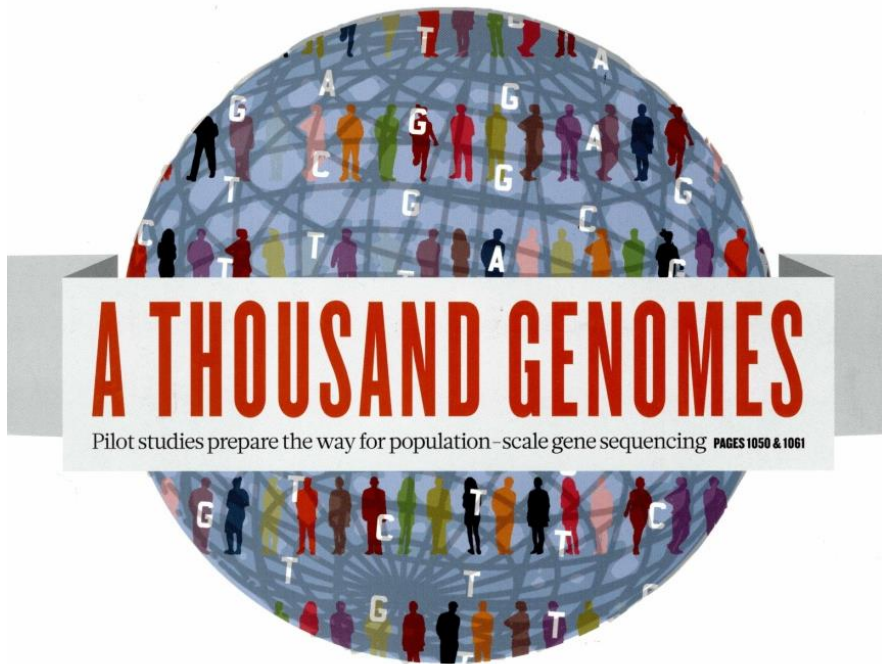
45 individuals from Beijing, China (CHB)

45 individuals from Tokyo, Japan (JPT)

The International HapMap Consortium (2005). A haplotype map of the human genome. *Nature*.

nature

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1000 Genomes Project

Phase 1: 1,092 individuals from 14 populations..

Phase 3: 2,504 individuals from 26 populations (~500 samples form each 5 continental ancestry groups, with ~5 populations for each group)

Population		Code	Population Color	Continental Group Color	Analysis Panel	Phase 1	Phase 3
African ancestry							
Esan in Nigeria	Esan	ESN			AFR		99
Gambian in Western Division, Mandinka	Gambian	GWD			AFR		113
Luhya in Webuye, Kenya	Luhya	LWK			AFR	97	99
Mende in Sierra Leone	Mende	MSL			AFR		85
Yoruba in Ibadan, Nigeria	Yoruba	YRI			AFR	88	108
African Caribbean in Barbados	Barbadian	ACB			AFR/AMR		96
People with African Ancestry in Southwest USA	African-American SW	ASW			AFR/AMR	61	61
Americas							
Colombians in Medellin, Colombia	Colombian	CLM			AMR	60	94
People with Mexican Ancestry in Los Angeles, CA, USA	Mexican-American	MXL			AMR	66	64
Peruvians in Lima, Peru	Peruvian	PEL			AMR		85
Puerto Ricans in Puerto Rico	Puerto Rican	PUR			AMR	55	104
East Asian ancestry							
Chinese Dai in Xishuangbanna, China	Dai Chinese	CDX			EAS		93
Han Chinese in Beijing, China	Han Chinese	CHB			EAS	97	103
Southern Han Chinese	Southern Han Chinese	CHS			EAS	100	105
Japanese in Tokyo, Japan	Japanese	JPT			EAS	89	104
Kinh in Ho Chi Minh City, Vietnam	Kinh Vietnamese	KHV			EAS		99
European ancestry							
Utah residents (CEPH) with Northern and Western European ancestry	CEPH	CEU			EUR	85	99
British in England and Scotland	British	GBR			EUR	89	91
Finnish in Finland	Finnish	FIN			EUR	93	99
Iberian Populations in Spain	Spanish	IBS			EUR	14	107
Toscani in Italia	Tuscan	TSI			EUR	98	107
South Asian ancestry							
Bengali in Bangladesh	Bengali	BEB			SAS		86
Gujarati Indians in Houston, TX, USA	Gujarati	GIH			SAS		103
Indian Telugu in the UK	Telugu	ITU			SAS		102
Punjabi in Lahore, Pakistan	Punjabi	PJL			SAS		96
Sri Lankan Tamil in the UK	Tamil	STU			SAS		102
Total						1092	2504

HUMAN STEM CELLS
BEYOND THE COURT CASE
 Implications for the law, industry and ethics
 PAGE 1031

OCEAN PRODUCTIVITY
PHOSPHATE DOWN THE AGES
 Key nutrient plentiful after 'snowball' Earth
 PAGES 1052 & 1080

AUTUMN BOOKS
THE RECURRING UNIVERSE
 Lee Smolin on Roger Penrose's grand idea
 PAGE 1034

NATURE.COM/NATURE
 28 October 2010 £10
 Vol. 467, No. 7319
 9 770028 083095

The 1000 Genomes Project Consortium (2012). An integrated map of genetic variation from 1,092 human genomes. *Nature*.

The 1000 Genomes Project Consortium (2015). A global reference for human genetic variation. *Nature*.

The Haplotype Reference Consortium (HRC)



A reference panel of 64,976 haplotypes for genotype imputation

nature

**Sequencing of 53,831 diverse genomes from
the NHLBI TOPMed Program**

Taliun, D., Harris, D.N., Kessler, M.D. *et al.* (2021). Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. *Nature*



Browse/Select Species

POPULAR SPECIES



Human



Mouse



Rat



Zebrafish



Fruitfly



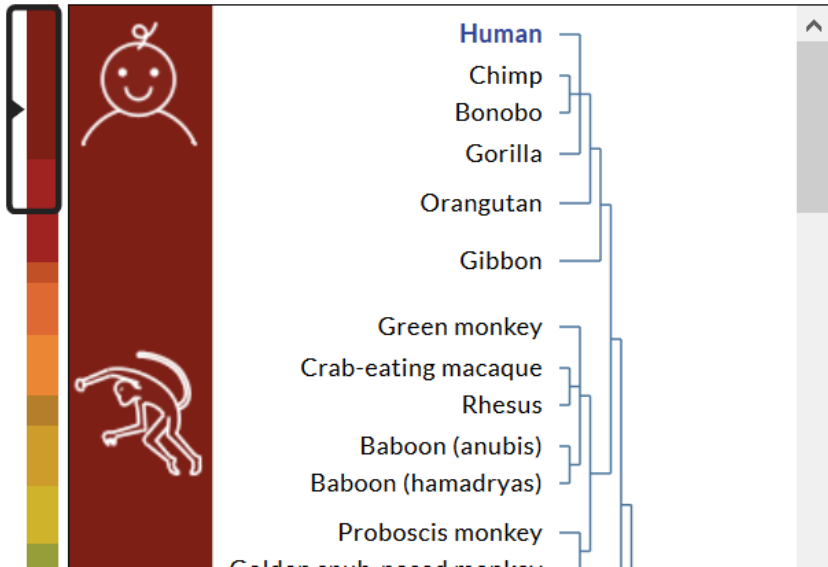
Worm



Yeast

[Can't find a genome assembly?](#)

REPRESENTED SPECIES



Find Position

Human Assembly



Position/Search Term

Current position: chrX:15,578,261-15,621,068

Human Genome Browser - hg19 assembly

[view sequences](#)

The February 2009 human reference sequence (GRCh37) was produced by the [Genome Reference Consortium](#). For more information about this assembly, see [GRCh37](#) in the NCBI Assembly database.

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

Request:

chr7

Genome Browser Response:

Displays all of chromosome 7



Homo sapiens
(Graphic courtesy of CBSE)

Assembly	Year
NCBI34/hg16	2003
NCBI35/hg17	2004
NCBI36/hg18	2006
GRCh37/hg19	2009
GRCh38/hg38	2014

NCBI / GRCh



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