A global reference for human genetic variation - DTU Orbit (03/12/2018)

A global reference for human genetic variation
The 1000 Genomes Project set out to provide a comprehensive description of common human genetic variation by applying whole-genome sequencing to a diverse set of individuals from multiple populations. Here we report completion of the project, having reconstructed the genomes of 2,504 individuals from 26 populations using a combination of low-coverage whole-genome sequencing, deep exome sequencing, and dense microarray genotyping. We characterized a broad spectrum of genetic variation, in total over 88 million variants (84.7 million single nucleotide polymorphisms (SNPs), 3.6 million short insertions/deletions (indels), and 60,000 structural variants), all phased onto high-quality haplotypes. This resource includes >99% of SNP variants with a frequency of >1% for a variety of ancestries. We describe the distribution of genetic variation across the global sample, and discuss the implications for common disease studies.

General information
State: Published
Organisations: Department of Systems Biology, Center for Biological Sequence Analysis, Albert Einstein College of Medicine of Yeshiva University, Vertex Pharmaceuticals, Wellcome Trust Sanger Institute, Chesterford Research Park, Johns Hopkins University, Cornell University, University of Oxford, University of Washington, University of Copenhagen, University of Michigan, European Bioinformatics Institute
Number of pages: 20
Pages: 68-74
Publication date: 2015
Peer-reviewed: Yes

Publication information
Journal: Nature
Volume: 526
ISSN (Print): 0028-0836
Ratings:
BFI (2018): BFI-level 3
Web of Science (2018): Indexed yes
BFI (2017): BFI-level 2
Scopus rating (2017): CiteScore 14.59
Web of Science (2017): Impact factor 19.181
Web of Science (2017): Indexed yes
BFI (2016): BFI-level 2
Scopus rating (2016): CiteScore 13.33
Web of Science (2016): Impact factor 19.304
Web of Science (2016): Indexed yes
BFI (2015): BFI-level 2
Scopus rating (2015): CiteScore 14.38
Web of Science (2015): Impact factor 17.184
Web of Science (2015): Indexed yes
BFI (2014): BFI-level 2
Scopus rating (2014): CiteScore 14.22
Web of Science (2014): Impact factor 14.547
Web of Science (2014): Indexed yes
BFI (2013): BFI-level 2
Scopus rating (2013): CiteScore 14.96
Web of Science (2013): Impact factor 15.295
ISI indexed (2013): ISI indexed yes
Web of Science (2013): Indexed yes
BFI (2012): BFI-level 2
Scopus rating (2012): CiteScore 14.01
ISI indexed (2012): ISI indexed yes
Web of Science (2012): Indexed yes
BFI (2011): BFI-level 2