

1.1 Some Very Useful Numbers for human population genetics.

When you're thinking about genomes and genomic data it's often useful to have a sense of rates and scales¹.

Genome properties.

Genome size: 3.1 Gb (haploid size).
Number of chromosomes: 23 pairs
Number of coding genes: ~20,000
Exons per gene: 8 (median)
Number of genes per megabase: 6.5 (mean)
Total in protein-coding exons: 1% of genome
Total in genes (introns+exons): 40% of genome
Active chromatin (per cell type): 1% of genome
Active chromatin (all cell types): 13% of genome

Length scales. (Orders of magnitude.)

Transcription factor binding site: 10 bp
Enhancer: 100 bp – 1 Kb
Exon (coding): 150 bp
Coding length per gene: 1200 bp (median)
Intron: 1 – 50 Kb
Gene (pre-mRNA): 10 – 100 Kb
Extent of LD: 10 Kb – 1 Mb (varies by locus & population)
Enhancer–promoter interactions: 1 Kb – 1 Mb
Chromatin topological domains (TADs): ~1 Mb
Chromosome lengths: 47 Mb – 250 Mb

Genetic variation.

Heterozygosity: $0.5\text{--}1.0 \times 10^{-3}$ (varies by population)
Human-Chimpanzee divergence: 1.4×10^{-2}
Number of common SNPs: ~8 million (at > 5% MAF in global sample)
Number of SNPs for genome-wide SNP tagging: ~1 million
Fst between populations: ~0.10–0.15 between continents

Population genetic parameters.

Mutation rate per generation: 1.3×10^{-8} per basepair (at parental age 30)
Mutation rate per year: 4.0×10^{-10} per basepair
Number of mutations per child: ~70
Recombination rate: 1.2 centiMorgans per Mb (mean, sex-averaged)
Cross-overs per egg: ~42
Cross-overs per sperm: ~26
Effective population size: 10,000–20,000 ($H/4\mu$)

Timescales of population divergence. (Take with grain of salt)

Human to chimpanzee: 6.5 MY
Human to Neanderthal/Denisovan: 600 KY
Deepest human population splits: ~200 KY
Out-of-Africa migration: 65–100 KY
Deepest non-African splits: 65 KY

An Owner's Guide to the Human Genome, by JK Pritchard. September 23, 2023. Original material distributed under a CC BY 4.0 license.

Notes and References.

¹Here's an excellent book-length treatment of this topic, with a focus on cell biology, free online [\[Link\]](#):
Milo R, Phillips R. Cell biology by the numbers. Garland Science; 2015