1000 Genomes

- Sequenciaram 1092 indivíduos de diversas populações.
- Identificação das variações genéticas encontradas.
- Frequências por população.
- Impacto.
- 2012

Table 1: GWAS for common diseases and traits

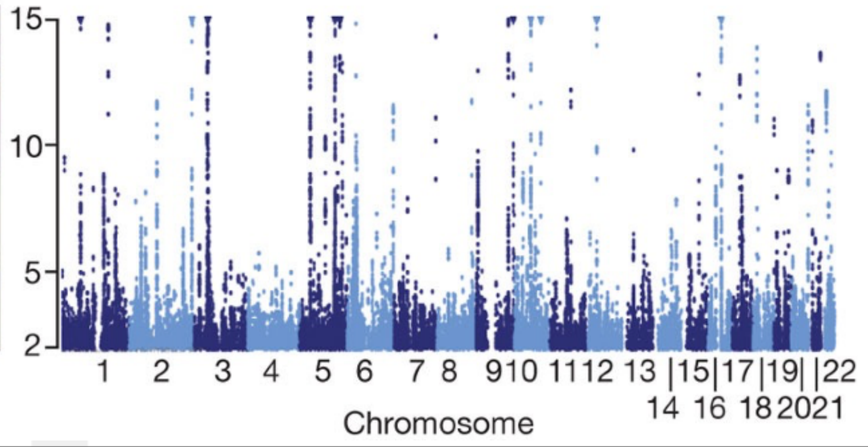
Phenotype	Number of GWAS loci	Proportion of heritability explained (%)*
Type 1 diabetes	41	~60
Fetal haemoglobin levels	3	~50
Macular degeneration	3	~50
Type 2 diabetes	39	20–25
Crohn's disease	71	20–25
LDL and HDL levels	95	20–25
Height	180	~12

ARTICLE

doi:10.1038/nature11632

An integrated map of genetic variation from 1,092 human genomes

The 1000 Genomes Project Consortium*



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An integrated map of structural variation in 2,504 human genomes

A list of authors and their affiliations appears at the end of the paper.

A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants

Advanced age-related macular degeneration (AMD) is the leading cause of blindness in the elderly, with limited therapeutic options. Here we report on a study of >12 million variants, including 163,714 directly genotyped, mostly rare, protein-altering variants. Analyzing 16,144 patients and 17,832 controls, we identify 52 independently associated common and rare variants ($P < 5 \times 10^{-8}$) distributed across 34 loci. Although wet and dry AMD subtypes exhibit predominantly shared genetics, we identify the first genetic association signal specific to wet AMD, near *MMP9* (difference *P* value = 4.1×10^{-10}). Very rare coding variants (frequency <0.1%) in *CFH*, *CFI* and *TIMP3* suggest causal roles for these genes, as does a splice variant in *SLC16A8*. Our results support the hypothesis that rare coding variants can pinpoint causal genes within known genetic loci and illustrate that applying the approach systematically to detect new loci requires extremely large sample sizes.