

THE SEQUENCE AND GENOTYPE VARIATION IN 150,119 GENOMES FROM THE UK BIOBANK

The relationship between sequences and phenotypic variation is usually obtained by exon sequencing, on the assumption that genes are much more important in this matter than the rest of the genome. However, [Stefansson et al.](#)¹ (Nature) observe that coding exons represent a small fraction of regions in the genome subject to strong sequence conservation. That means that they are evolutionarily important. The authors analyzed the high-quality whole genome of 150,119 individuals in the UK biobank, and identified 895,055 structural variants and 2,536,688 microsatellites typically excluded from large-scale whole-genome sequencing studies. This analysis discovered “several examples of trait associations for rare variants with large effects not found previously through studies based on whole-exome sequencing and/or imputation”.

1-<https://www.nature.com/articles/s41586-022-04965-x>