Genome Trax Statistics

TRACK NAME	SOURCE & VERSION	TOTAL (HG38)
Mutations and variants		
HGMD® inherited disease mutations	HGMD® Professional 2020.2	260,703
HGMD® imputed inherited disease mutations	HGMD® Professional 2020.2	30,029
PGMD® pharmacogenomics variants (Beta)	PGMD® 2016.2 (beta)	1,108,238
ClinVar Variants	ClinVar-2020-06	1,019,044
GWAS Catalogue	Downloaded on 20th Jun 2020	180,007
EVS Exome Variations	ESP6500SI-V2	3,686,725
dbNSFP Nonsynonymous functional predictions	v3.5c	83,305,895
dbSNP	dbSNP150	325,000,482
Allele frequencies from 1000 Genomes (Beta)	ENSEMBL 99	12,237,697
TCGA	Downloaded on 12th Jun 2016	54,74,701
Complete Genomics Allele Frequency	Downloaded on 15th Sep 2015	25,110,354
Regulatory Features		
TRANSFAC® experimentally verified TFBS	TRANSFAC® 2020.2	64,397
Predicted ChIP-Seq TFBS	TRANSFAC® 2020.2	44,387,408
Predicted TFBSs in DNAse hypersensitivity regions	TRANSFAC® 2020.2	10,815,746
CpG Islands	TRANSFAC® 2020.2	48,300
Microsatellites	ENSEMBL 99	1,405,943
Virtual Transcription Start Sites (TSSs)	TRANSFAC® 2020.2	88,492
Post translational modifications	PROTEOME™ 2020.2	53,129
miRNA	miRBase 21	2,813
dbscSNV Variations in splice sites	v1.1	15,027,837
Vista Enhancers	Downloaded on Jun 2020	1,921
Gene Functional Assignments		
Disease genes	PROTEOME™ 2020.2	14,203
Drug targets	PROTEOME™ 2020.2	2,914
Pathway membership	PROTEOME™ 2020.2	1,323
HGMD® disease associations	HGMD® Professional 2020.2	49,536
Orphanet	Downloaded on 1st Aug 2016	6,482
OMIM	Downloaded on 24th May 2017	3,669

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