

NHGRI GWAS catalog: description of column headings for Tab delimited File

Note: The SNP data in the catalog has been mapped to dbSNP Build 141 and Genome Assembly, GRCh38/hg38.

DATE ADDED TO CATALOG: Date added to catalog
PUBMEDID: PubMed identification number
FIRST AUTHOR: Last name of first author
DATE: Publication date (online (epub) date if available)
JOURNAL: Abbreviated journal name
LINK: PubMed URL
STUDY: Title of paper (linked to PubMed abstract)
DISEASE/TRAIT: Disease or trait examined in study
INITIAL SAMPLE SIZE: Sample size for Stage 1 of GWAS
REPLICATION SAMPLE SIZE: Sample size for subsequent replication(s)
REGION: Cytogenetic region associated with rs number (NCBI)
CHR_ID: Chromosome number associated with rs number (NCBI)
CHR_POS: Chromosomal position associated with rs number (dbSNP Build 132, NCBI)
REPORTED GENE (S): Gene(s) reported by author
MAPPED GENE(S): Gene(s) mapped to the strongest SNP (NCBI). If the SNP is located within a gene, that gene is listed. If the SNP is intergenic, the upstream and downstream genes are listed, separated by a hyphen. UPSTREAM_GENE_ID: Entrez Gene ID for nearest upstream gene to rs number, if not within gene (NCBI)
DOWNSTREAM_GENE_ID: Entrez Gene ID for nearest downstream gene to rs number, if not within gene (NCBI)
SNP_GENE_IDS: Entrez Gene ID, if rs number within gene; multiple genes denotes overlapping transcripts (NCBI)
UPSTREAM_GENE_DISTANCE: distance in kb for nearest upstream gene to rs number, if not within gene (NCBI)
DOWNSTREAM_GENE_DISTANCE: distance in kb for nearest downstream gene to rs number, if not within gene (NCBI)
STRONGEST SNP-RISK ALLELE: SNP(s) most strongly associated with trait + risk allele (? for unknown risk allele). May also refer to a haplotype.
SNPS: Strongest SNP; if a haplotype is reported above, may include more than one rs number (multiple SNPs comprising the haplotype)
MERGED: denotes whether the SNP has been merged into a subsequent rs record (0 = no; 1 = yes; NCBI)
SNP_ID_CURRENT: current rs number (will differ from strongest SNP when merged = 1)
CONTEXT: SNP functional class (NCBI)
INTERGENIC: denotes whether SNP is in intergenic region (0 = no; 1 = yes; NCBI)
RISK ALLELE FREQUENCY: Reported risk allele frequency associated with strongest SNP

P-VALUE: Reported p-value for strongest SNP risk allele (linked to dbGaP Association Browser)

PVALUE_MLOG: $-\log(\text{p-value})$

P-VALUE (TEXT): Information describing context of p-value (e.g. females, smokers).

Note that p-values are rounded to 1 significant digit (for example, a published p-value of 4.8×10^{-7} is rounded to 5×10^{-7}).

OR or BETA: Reported odds ratio or beta-coefficient associated with strongest SNP risk allele

95% CI (TEXT): Reported 95% confidence interval associated with strongest SNP risk allele

PLATFORM (SNPS PASSING QC): Genotyping platform manufacturer used in Stage 1; also includes notation of pooled DNA study design or imputation of SNPs, where applicable

CNV: Study of copy number variation (yes/no)

Updated: September 18, 2014