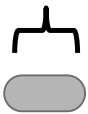
SNP

Identifier: RS number, example “rs1234”.
Used to refer to a known and documented SNP whose position can be retrieved from the knowledge database.

Position

Identifier: Chromosome and basepair location, example “chr1:234”.
Used to refer to any single genomic location, such as a single nucleotide polymorphism (SNP), single nucleotide variation (SNV), rare variant, or any other position of interest.

Region



Identifier: Chromosome and basepair range, example “chr1:234-567”.
Used to refer to any genomic region, such as a copy number variation (CNV), insertion/deletion (indel), gene coding region, evolutionarily conserved region (ECR), functional region, regulatory region, or any other region of interest.

Gene

Identifier: Name, example “A1BG” or “ENSG00000121410”.
Used to refer to a known and documented gene, whose genomic region and associations with any pathways, interactions or other groups can be retrieved from the knowledge database.

Group

Identifier: Name, example i.e. “lipid metabolic process” or “GO:0006629”.
Used to refer to a known and documented pathway, ontological group, protein interaction, protein family, or any other grouping of genes, proteins or genomic regions that was provided by one of the external data sources.

Source

Identifier: Name, example “GO”.
Used to refer to a specific external data source.