

Import of Human GWAS Data and Mapping of EFO to multiple ontologies at RGD

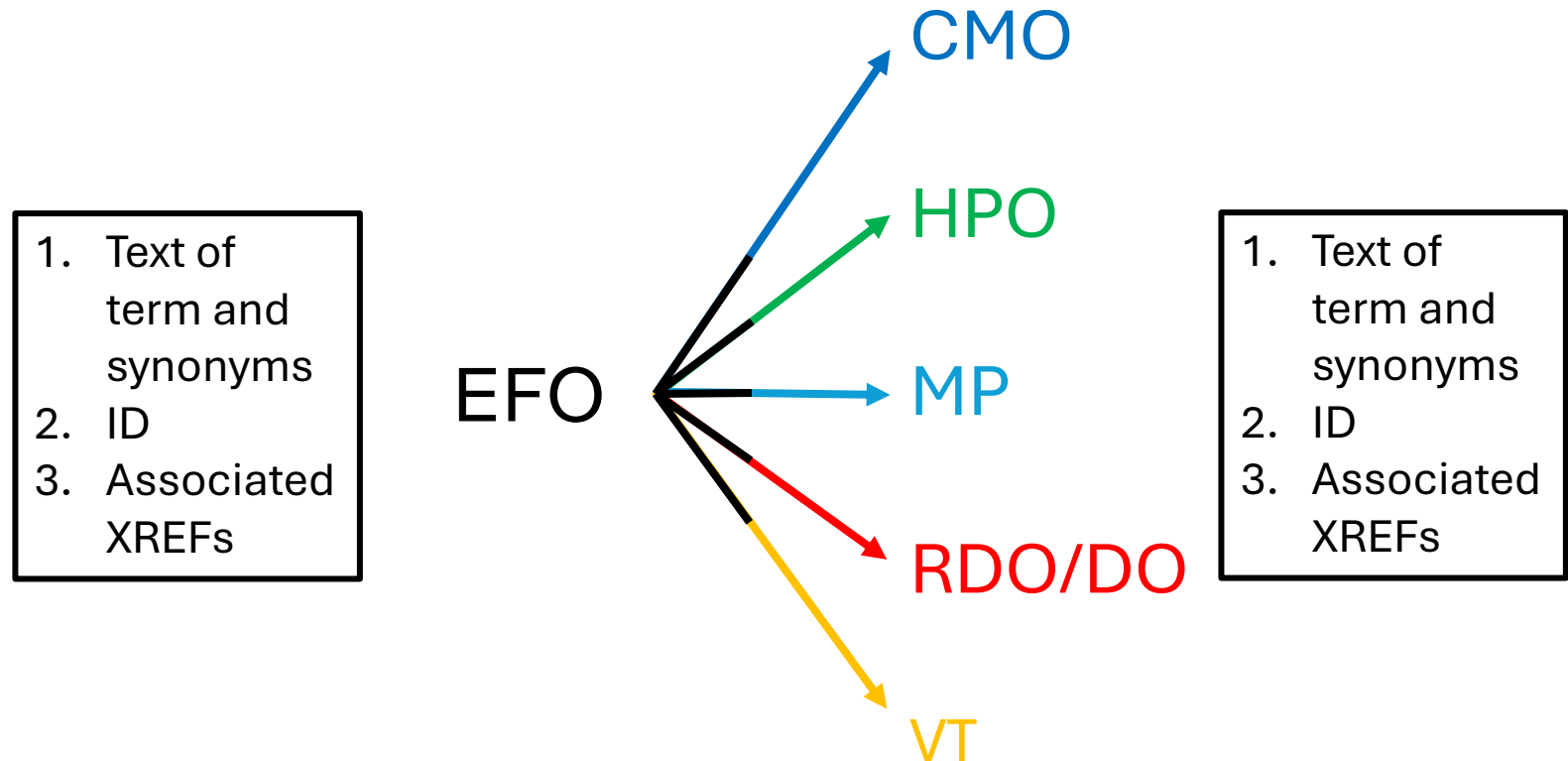
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Rat Genome Database
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Biocuration 2025

Ontologies used to curate phenotype and disease at the Rat Genome database (RGD)

Data type at RGD	Ontology used at RGD
rat phenotype (qualitative)	Mammalian Phenotype ontology (MP)
rat phenotype (quantitative)	Vertebrate Trait ontology (VT)
	Clinical Measurement Ontology (CMO)
	Measurement Method Ontology (MMO)
	Experimental Condition Ontology (XCO)
	Uberon ontology (UBERON)
	Cell Ontology (CL)
human phenotype (qualitative)	Human Phenotype Ontology (HPO)
disease (qualitative)	RGD disease ontology (RDO) (axiomatized and extended version of the human disease ontology (DO))



Automated term mapping



Text of EFO/target
term

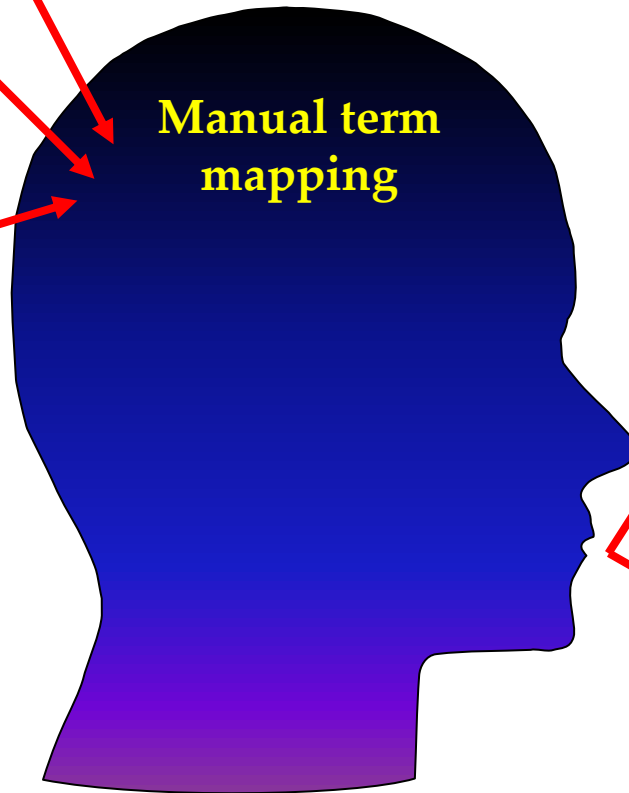
Definition of
EFO/target term

Synonyms of
EFO/target term

**Manual term
mapping**

Text of EFO term
added to target
term as synonym
(exact, broad,
narrow or related)

ID of EFO term
added to target
term as XREF



Number of EFO terms from GWAS records mapped to ontologies at RGD

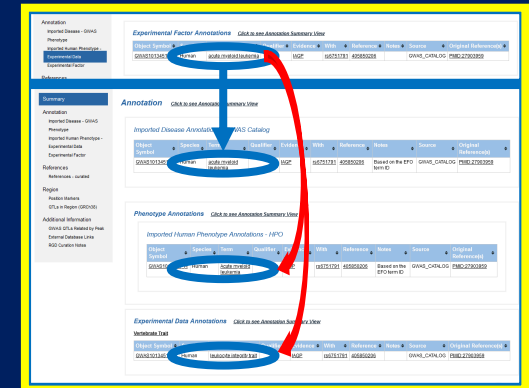
EFO term subsets	Subset Numbers as of March 2025
total # of unique EFO terms associated with imported GWAS records	7601
total # of unique EFO/HP/MONDO hybrid terms associated with imported GWAS records	2009
# of EFO terms mapped to RDO/DO	1930
# of EFO terms mapped to HPO	514
# of EFO terms mapped to CMO	469
# of EFO terms mapped to MP	209
# of EFO terms mapped to VT	900+

Term mapping output for users

1. Entries in term browser (HPO, CMO, MP, RDO/DO, and VT)










2. Transitive annotations on Human QTL pages (SNP variant pages)



3. SSSOM file on the RGD download page
(<https://download.rgd.mcw.edu/ontology/mappings/>)

Index of /ontology/mappings

Name	Last modified	Size	Description
 Parent Directory			
 efo_cmo_rgd.sssom.tsv.txt	2025-03-22 19:14	52K	
 efo_hp_rgd.sssom.tsv.txt	2025-03-22 19:14	126K	
 efo_mp_rgd.sssom.tsv.txt	2025-03-22 19:14	26K	
 efo_rdo_rgd.sssom.tsv.txt	2025-03-22 19:14	1.3M	
 efo_vt_rgd.sssom.tsv.txt	2025-03-22 19:14	111K	
 rdo_mim_rgd.sssom.tsv.txt	2025-03-22 19:14	1.5M	

EFO entry in term browser: acute myeloid leukemia

acute myeloid leukemia (EFO:0000222)

Annotations: Rat: (0) Mouse: (0) Human: (1744) Chinchilla: (0) Bonobo: (0) Dog: (0) Squirrel: (0) Pig: (0) Naked Mole-rat: (0) Green Monkey: (0)

Parent Terms	Term With Siblings	Child Terms
<p>Acute Leukemia + </p> <p>myeloid leukemia + </p>	<p>acute lymphoblastic leukemia + </p> <div style="border: 1px solid black; padding: 5px; margin: 5px 0;"> <p>acute myeloid leukemia + </p> <p>Acute myeloid leukemia (AML) is a group of neoplasms arising from precursor cells committed to the myeloid cell-line differentiation. All of them are characterized by clonal expansion of myeloid blasts. AML manifests by fever, pallor, anemia, hemorrhages and recurrent infections.</p> </div> <p>atypical chronic myeloid leukemia, BCR-ABL1 negative </p> <p>chronic myelogenous leukemia + </p> <p>leukemia, acute, X-linked </p>	<p> acute erythroblastic leukemia </p> <p> acute leukemia of ambiguous lineage + </p> <p> acute monocytic leukemia + </p> <p> acute myeloid leukemia by FAB classification + </p> <p> acute myeloid leukemia with CEBPA somatic mutations </p> <p> acute myeloid leukemia with multilineage dysplasia </p> <p> acute myeloid leukemia with mutated NPM1 </p> <p> acute myeloid leukemia, biallelic CEBPA gene mutation </p> <p> acute myeloid leukemia, CEBPA gene mutation </p> <p> acute myeloid leukemia, del(13q14-q21) </p> <p> acute myeloid leukemia, del(5q31-q32) </p> <p> acute myeloid leukemia, der12p </p> <p> acute myeloid leukemia, FLT3 internal tandem duplication </p> <p> acute myeloid leukemia, FLT3 tyrosine kinase domain point mutation </p>

Commonly used browser icons: is_a part_of regulates

Navigation: View annotations Pathway diagram (this term) Pathway diagram (child term(s))

Synonyms

Exact Synonyms: AML ; ANLL ; acute Nonlymphocytic leukaemia ; acute Nonlymphocytic leukemia ; acute granulocytic leukaemia ; acute granulocytic leukemia ; acute myeloblastic leukaemia ; acute myeloblastic leukemia ; acute myelocytic leukaemia ; acute myelocytic leukemia

Related Synonyms: acute non lymphoblastic leukaemia ; acute non lymphoblastic leukemia ; leukemia, acute myelogenous ; leukemia, acute myeloid, susceptibility to

Xrefs: DOID:9119 ; GARD:12757 ; GARD:537 ; HP:0004808 ICD10:C92.0 ; ICD10:C92.00 ; ICD9:205.0 ; ICD9:205.00 ; ICDO:9861/3 ; KEGG:05221

Transitive annotations on Human QTL pages (SNP variant pages)

The screenshot displays a web interface for a Human QTL page, specifically for the SNP variant rs6751791. The interface is divided into several sections, with a left sidebar containing navigation links. The main content area shows three tables of annotations, each with a title and a link to 'Click to see Annotation Summary View'.

Left Sidebar:

- Annotation
 - Imported Disease - GWAS
 - Phenotype
 - Imported Human Phenotype -
 - Experimental Data**
 - Experimental Factor
- References
 - References - curated
- Region
 - Position Markers
 - QTLs in Region (GRCh38)
- Additional Information
 - GWAS QTLs Related by Peak
 - External Database Links
 - RGD Curation Notes

Experimental Factor Annotations

Object Symbol	Species	Term	Qualifier	Evidence	With	Reference	Notes	Source	Original Reference(s)
GWAS1013451	Human	acute myeloid leukemia		IAGP	rs6751791	405850206		GWAS_CATALOG	PMID:27903959

Annotation

Imported Disease Annotations - GWAS Catalog

Object Symbol	Species	Term	Qualifier	Evidence	With	Reference	Notes	Source	Original Reference(s)
GWAS1013451	Human	acute myeloid leukemia		IAGP	rs6751791	405850206	Based on the EFO term ID	GWAS_CATALOG	PMID:27903959

Phenotype Annotations

Imported Human Phenotype Annotations - HPO

Object Symbol	Species	Term	Qualifier	Evidence	With	Reference	Notes	Source	Original Reference(s)
GWAS1013451	Human	Acute myeloid leukemia		IAGP	rs6751791	405850206	Based on the EFO term ID	GWAS_CATALOG	PMID:27903959

Experimental Data Annotations








Vertebrate Trait

Object Symbol	Species	Term	Qualifier	Evidence	With	Reference	Notes	Source	Original Reference(s)
GWAS1013451	Human	leukocyte integrity trait		IAGP	rs6751791	405850206		GWAS_CATALOG	PMID:27903959

Red arrows indicate the transitive flow of annotations: from 'acute myeloid leukemia' in the Experimental Factor table to 'acute myeloid leukemia' in the Imported Disease table, then to 'Acute myeloid leukemia' in the Imported Human Phenotype table, and finally to 'leukocyte integrity trait' in the Experimental Data table.

SSSOM files on the RGD download page

Index of /ontology/mappings

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Acknowledgements



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Rat Genome Database

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