

Mapping of EFO terms to multiple ontologies at RGD

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Abstract

The laboratory rat, Rattus norvegicus, is an important model of human health and disease, and experimental findings in the rat have relevance to human physiology and disease. The Rat Genome Database (RGD, https://rgd.mcw.edu) is a model organism database that provides access to a wide variety of curated rat data including disease associations, phenotypes, pathways, molecular functions, biological processes, cellular components, and chemical interactions for genes, quantitative trait loci, and strains. A major role of a model organism database is to provide data from laboratory animals in comparison to human data.

To be able to compare data between rat and human it is necessary to use common vocabularies. When data from different sources use different vocabularies, there must be some standard way to compare the data. To bridge gaps in data description from different sources, the biocurators at the Rat Genome Database (RGD) have mapped a human GWAS-specific subset of terms (The NHGRI-EBI Catalog of human genome-wide association studies/GWAS Catalog Data) (https://www.ebi.ac.uk/gwas/) from EFO (Experimental Factor Ontology) to multiple ontologies used at RGD. RGD has also made available the ontological mappings via SSSOM files on the RGD download site (https://download.rgd.mcw.edu/ontology/mappings/).

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Figure 3. Multiple browser views showing connections among EFO, RDO, HP, and MP terms. Red arrows show term-synonym relationships and term ID-XREF relationships. Blue highlights indicate that the selected terms have annotations (A) in RGD.

Hypertension resistant to conventional therapy (HP:0430034) Annotations: Rat: (0) Mouse: (0) Human: (2) Chinchilla: (0) Bonobo: (0) Dog: (0) Squirrel: (0) Pig: (0) Naked Mole-rat: (0) Green Monkey: (0)				
Pare	ent Terms	Term With Siblings	Child Terms	
Hypertension + A 分 II		Episodic hypertension Hypertension associated with pheochromocytoma + Hypertension resistant to conventional therapy Resistant hypertension is defined as above-goal elevated blood pressure in a patient despite the concurrent use of 3 antihypertensive drug classes, commonly including a long-acting calcium channel blocker, a blocker of the reninangiotensin system (angiotensin-converting enzyme infiliator or angiotensin receptor blocker), and a diuretic. Hypertensive crisis Hypertension Renovascular hypertension Renovascular hypertension Hypertensio		
Commonly used browser icor Term Relationships: I is_a	ns: ❷part_of ⊠regulates Navig	ation: 🔼 View annotations 🔟 Pathway diagram (this term) 🗯 Pathway diagram (child	d term(s))	
Svnonvms Exact Synonyms:	Registent hypertensies :	reatment registent hypertension		
Xrefs:	EFO:1002006	reatment-resistant hypertension		
xreis:	LI 0.1002000			

hypertension + A & I

Synonyms

coronary artery vasospasm + A & I

hypertension + A & I

Commonly used browser icons:

Term Relationships: ■ is_a part_of regulates

treatment-resistant hypertension

MESH:C563514 EFO:1002006

chemotherapy-induced hypertension A &

hypertension, pregnancy-incuced + A &

early onset hypertension A essential hypertension + A

intracranial hypertensi malignant hypertensio nephrosclerosis &

ocular hypertension [A]

secondary hypertens

Annotations: Rat: (3) Mouse: (3) Human: (81) Chinchilla: (2) Bonobo

xacerbation in Pregnancy 🔼 🚜

Isolated Systolic Hypertension A &

Pregnancy-Induced Hypertension + A &

hypertensive retinopathy 🔼 🙈

Kallikrein Hypertension &

Prinzmetal angina A

malignant hypertension + A Masked Hypertension A ocular hypertension + A &

pulmonary hypertension + A &

ertension, Early-Onset, Autosomal Dominant, with Severe

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

Hypomagnesemia, Hypertension, and Hypercholesterolemia

Idiopathic Intracranial Hypertension with Papilledema &

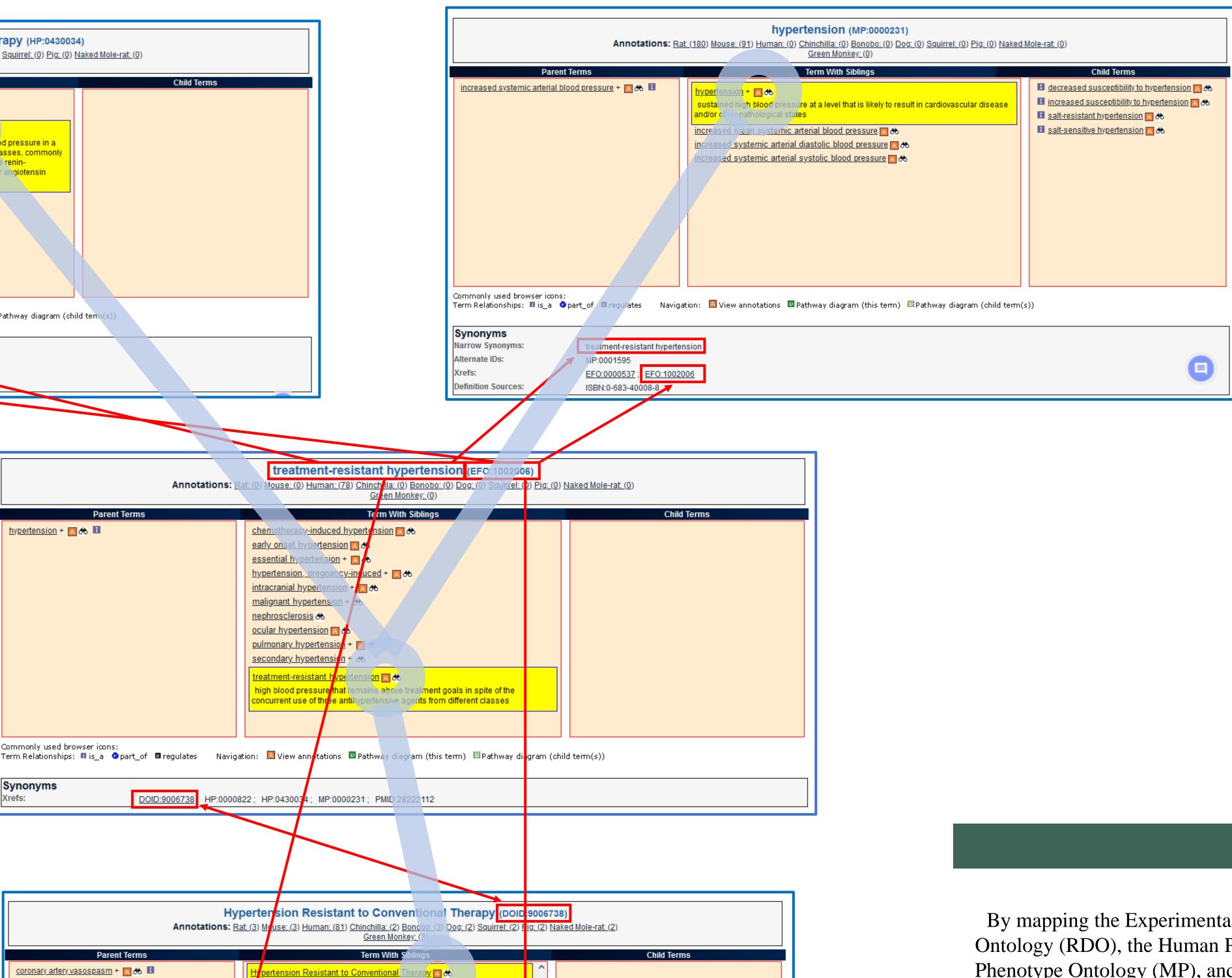
pulmonary hypertension + M

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)
	RGD disease ontology (RDO) (axiomatized
	and extended version of the human
disease (qualitative)	disease ontology (DO)

Figure 1. Ontologies used to curate phenotype and disease at the Rat Genome Database

EFO term subsets	Subset Numbers as of July 2024
otal # of unique EFO terms associated with imported GWAS	
records	760
otal # of unique EFO/HP/MONDO hybrid terms associated	
with imported GWAS records	200
of EFO terms mapped to RDO/DO	593
of EFO terms mapped to CMO	47
of EFO terms mapped to HPO	51
of EFO terms mapped to MP	20

Figure 2. EFO terms from GWAS records mapped to ontologies at RGD



Summary

By mapping the Experimental Factor Ontology (EFO) to the RGD Disease Ontology (RDO), the Human Phenotype Ontology (HPO), the Mammalian Phenotype Ontology (MP), and the Clinical Measurement Ontology (CMO), RGD has expanded the ability of database users to compare rat and other model organism data to human data and the reverse. The mapping of EFO terms to these various other ontologies will be continuously updated as more human GWAS data is imported to RGD. The ontology mappings will be a useful and necessary aide in translational research moving forward.