A semantic query interface for the OGO platform

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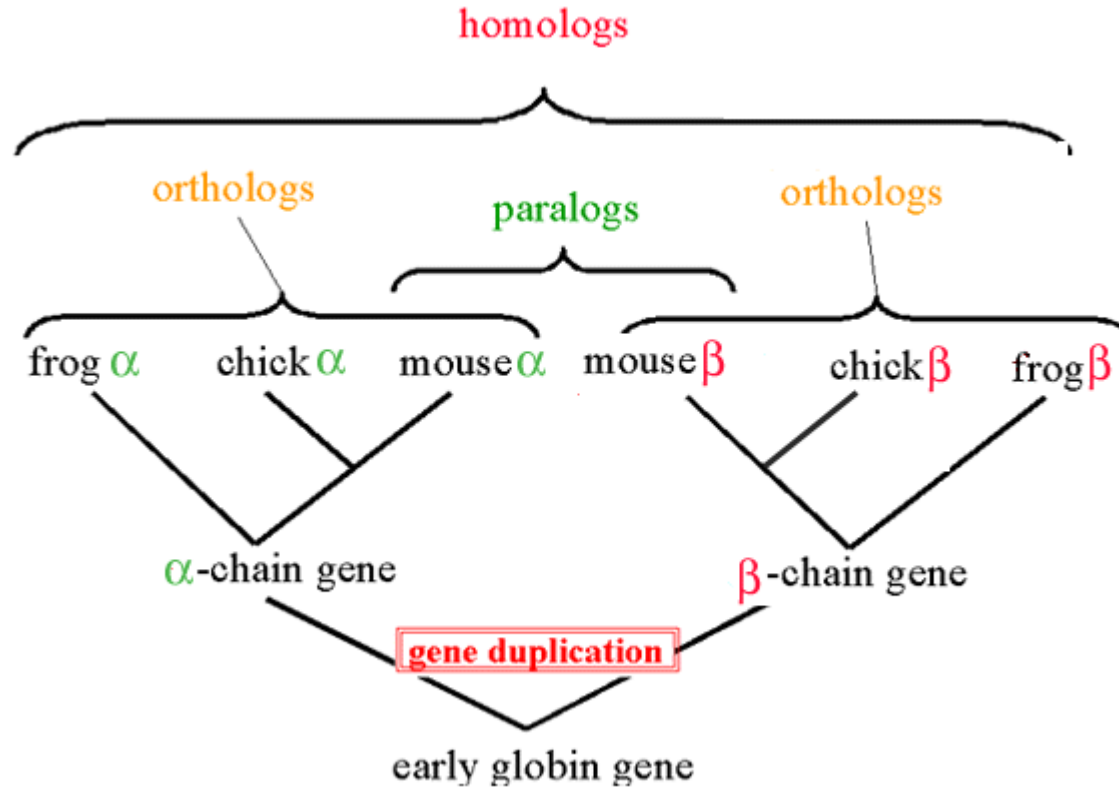http://tinyurl.com/35amhn6

Overview

Orthologs

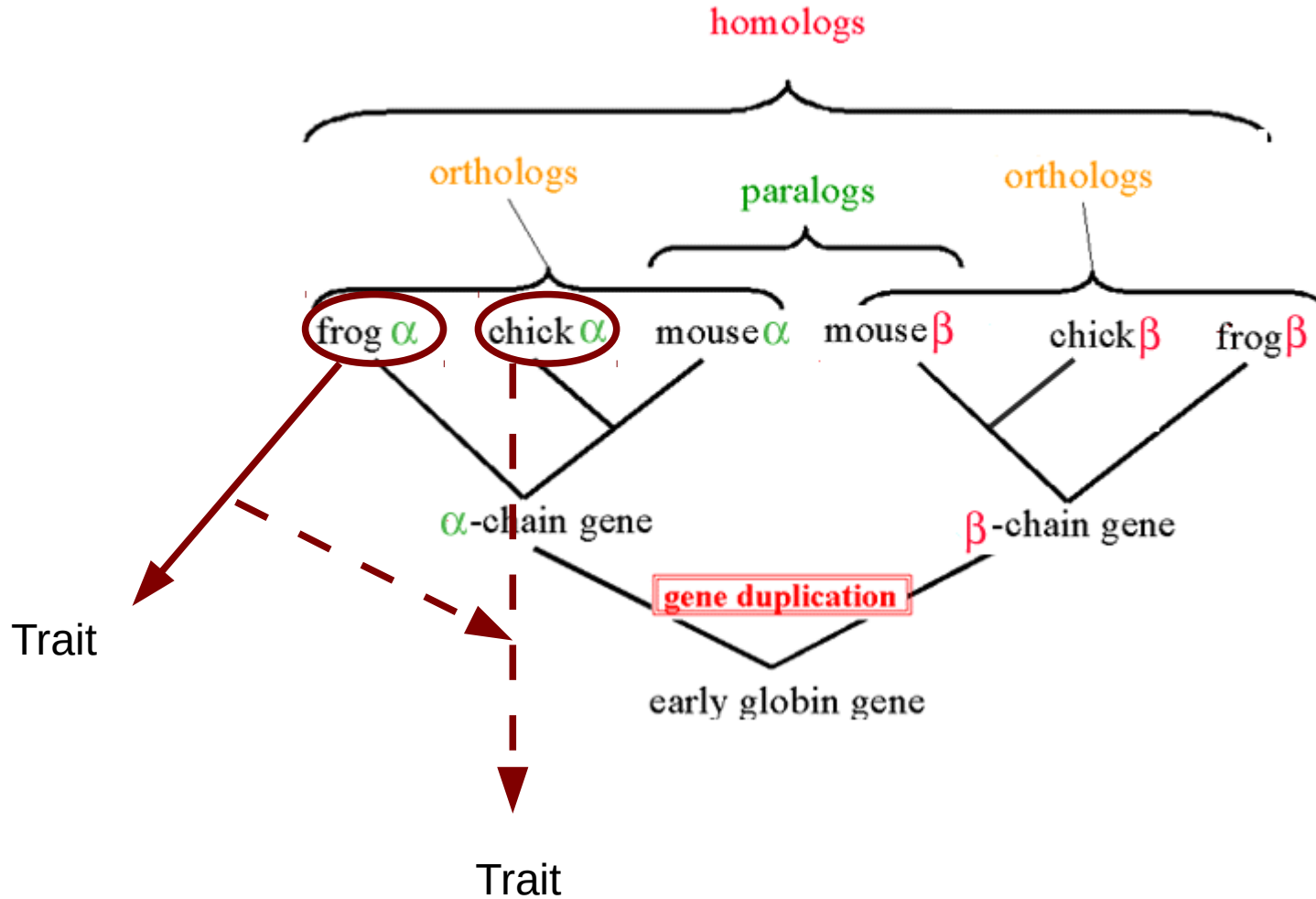Information about orthologs and diseases

OGO system

A semantic query interface for the OGO system

Sample query

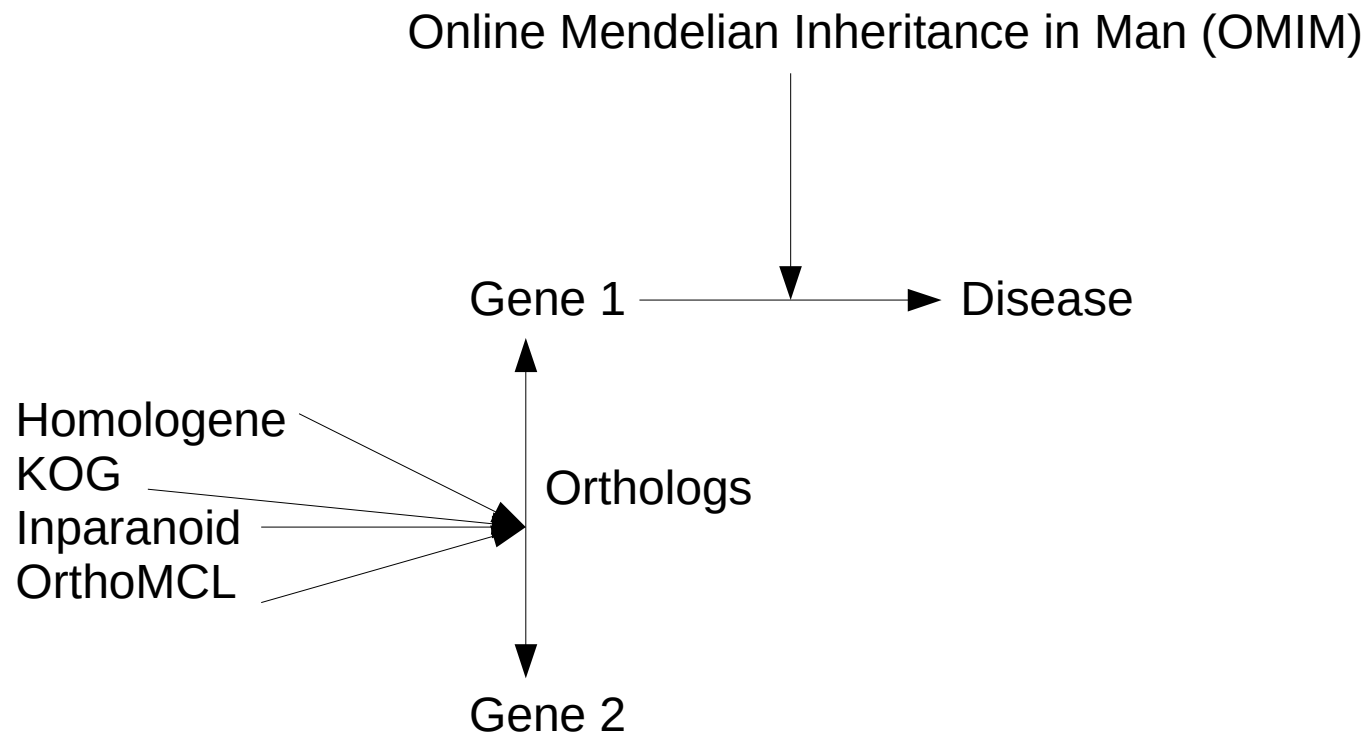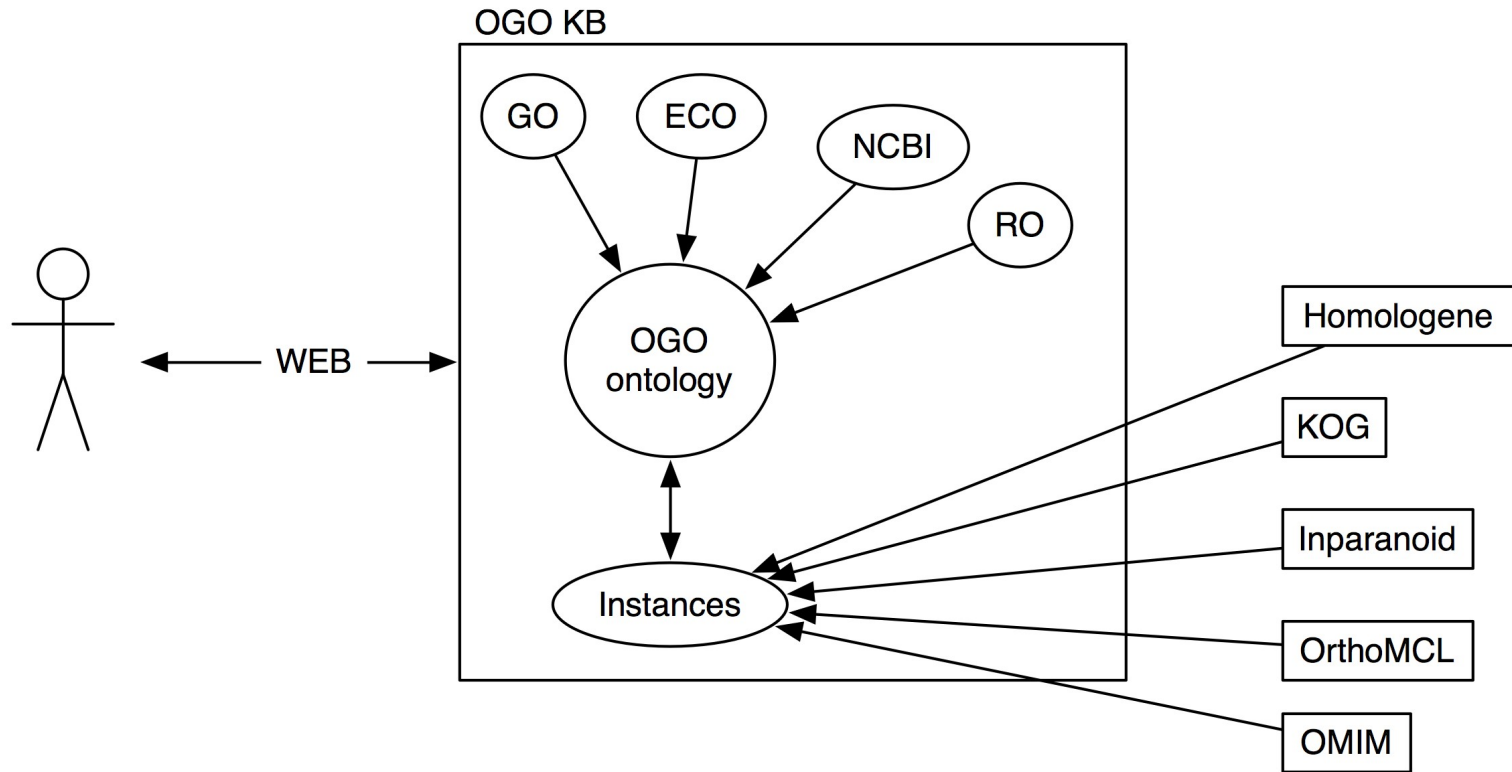# Ortholog sequences

Ortholog sequences

Orthologs and genetic diseases

Online Mendelian Inheritance in Man (OMIM)
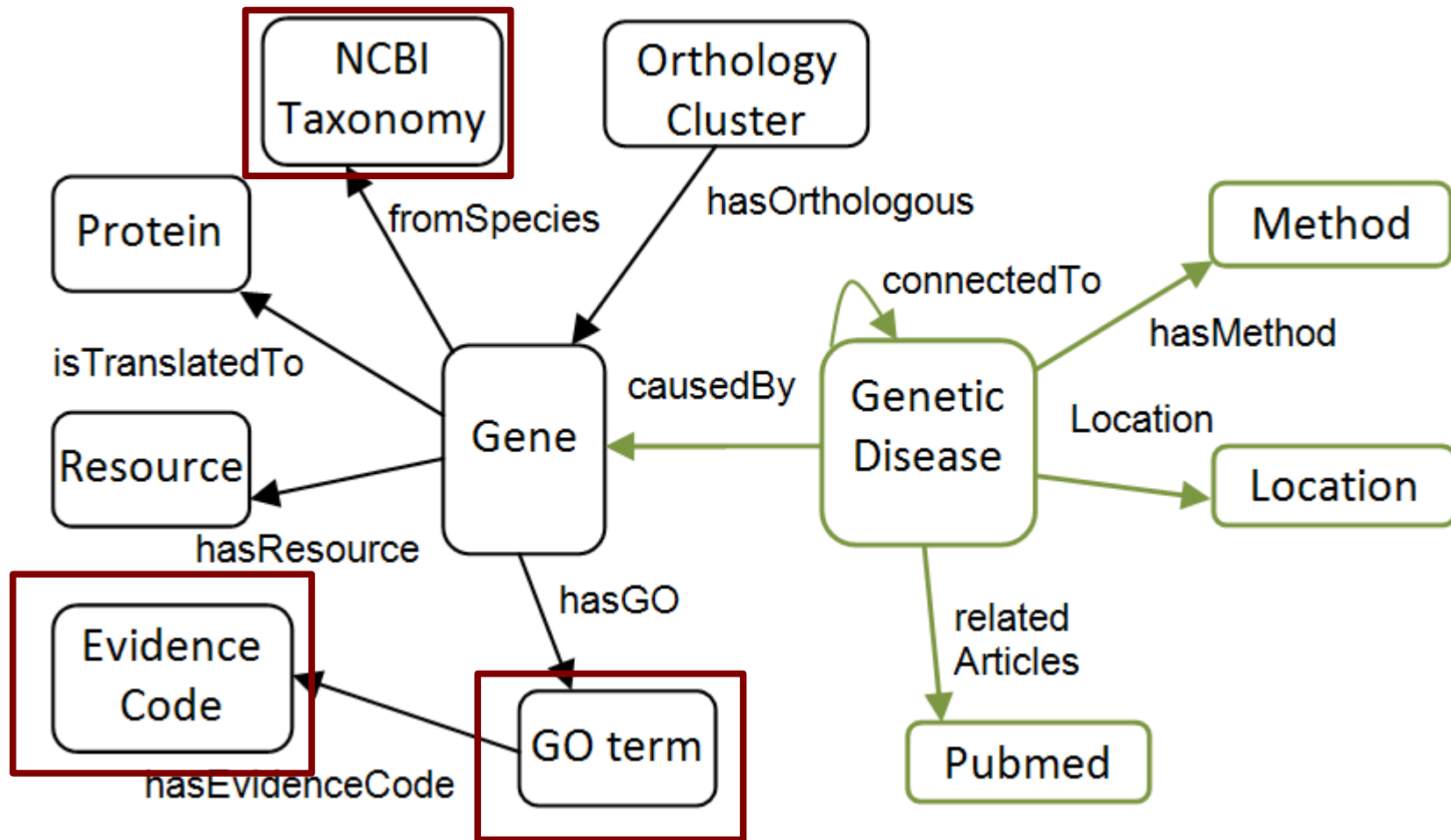
Gene 1 ⟶ Disease

Homologene
KOG
Inparanoid
OrthoMCL

Orthologs

Gene 2

OGO system

OGO ontology

OGO ontology: imported ontologies

Gene Ontology (OBOF): molecular function, biological process and cellular component of gene products

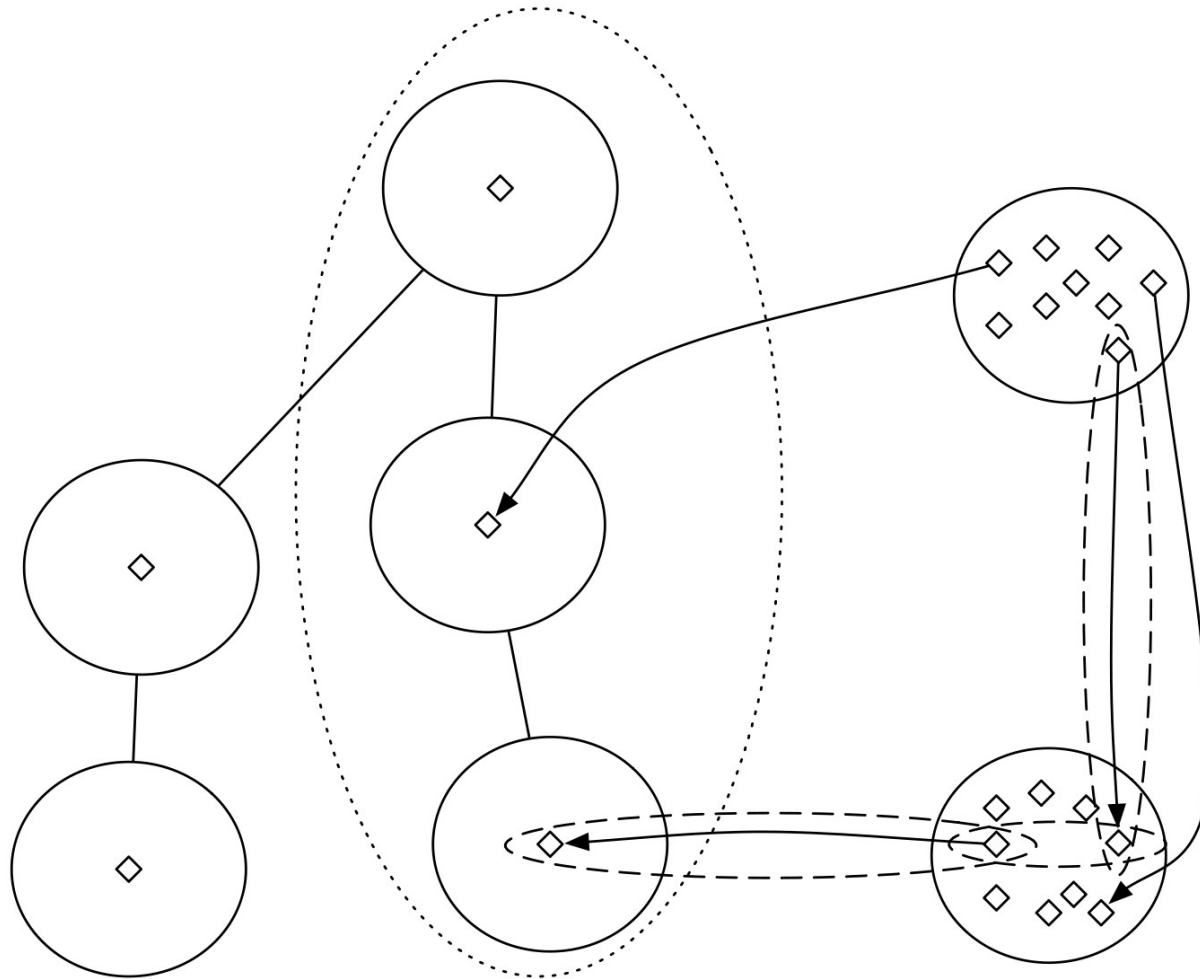Evidence Codes Ontology (Candidate OBOF): GO annotations evidence codes

OBO Relationship Types (Candidate OBOF):

```
Gene product participates in some (molecular function
    or biological process)
```
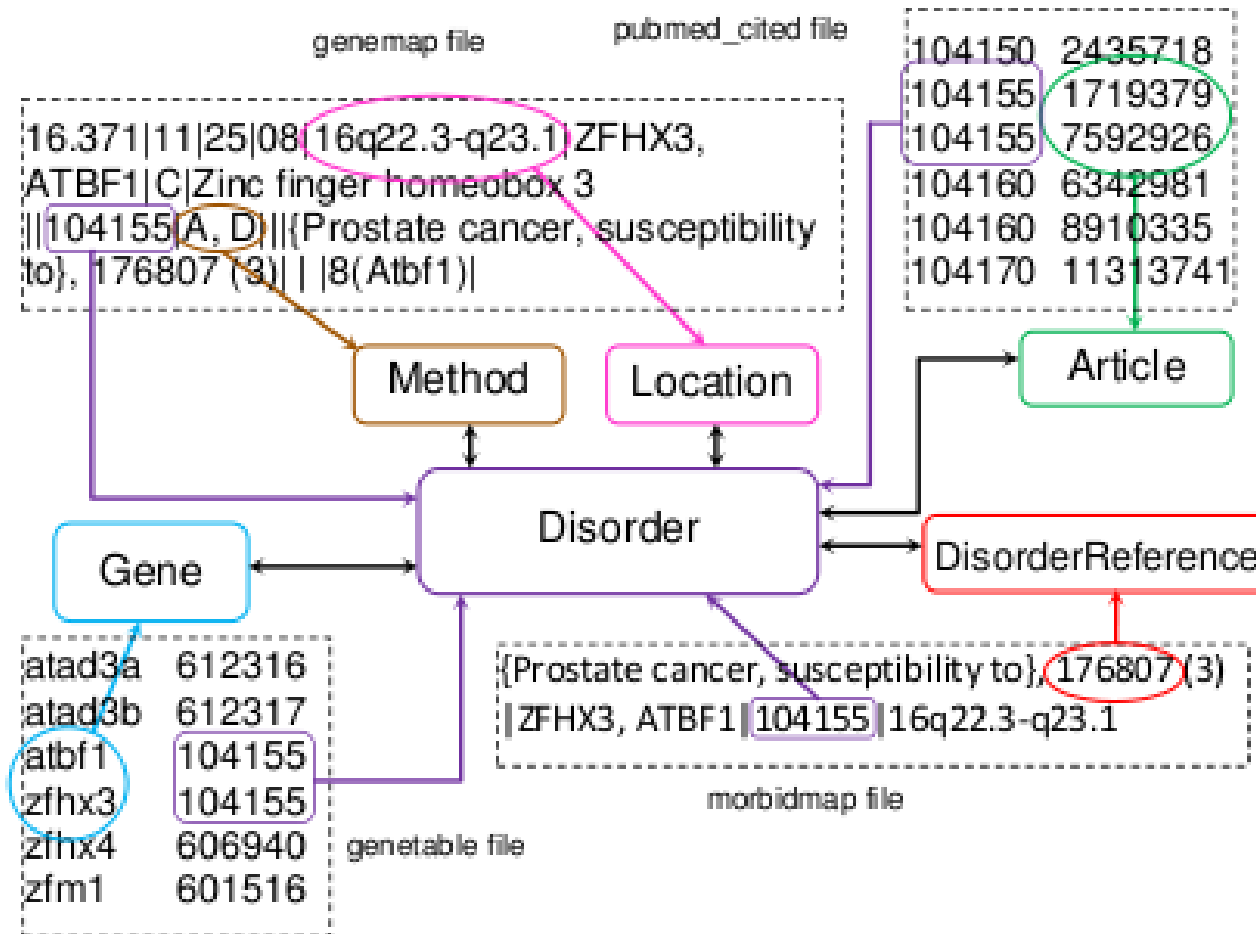
```
Gene product located in some cellular component
```
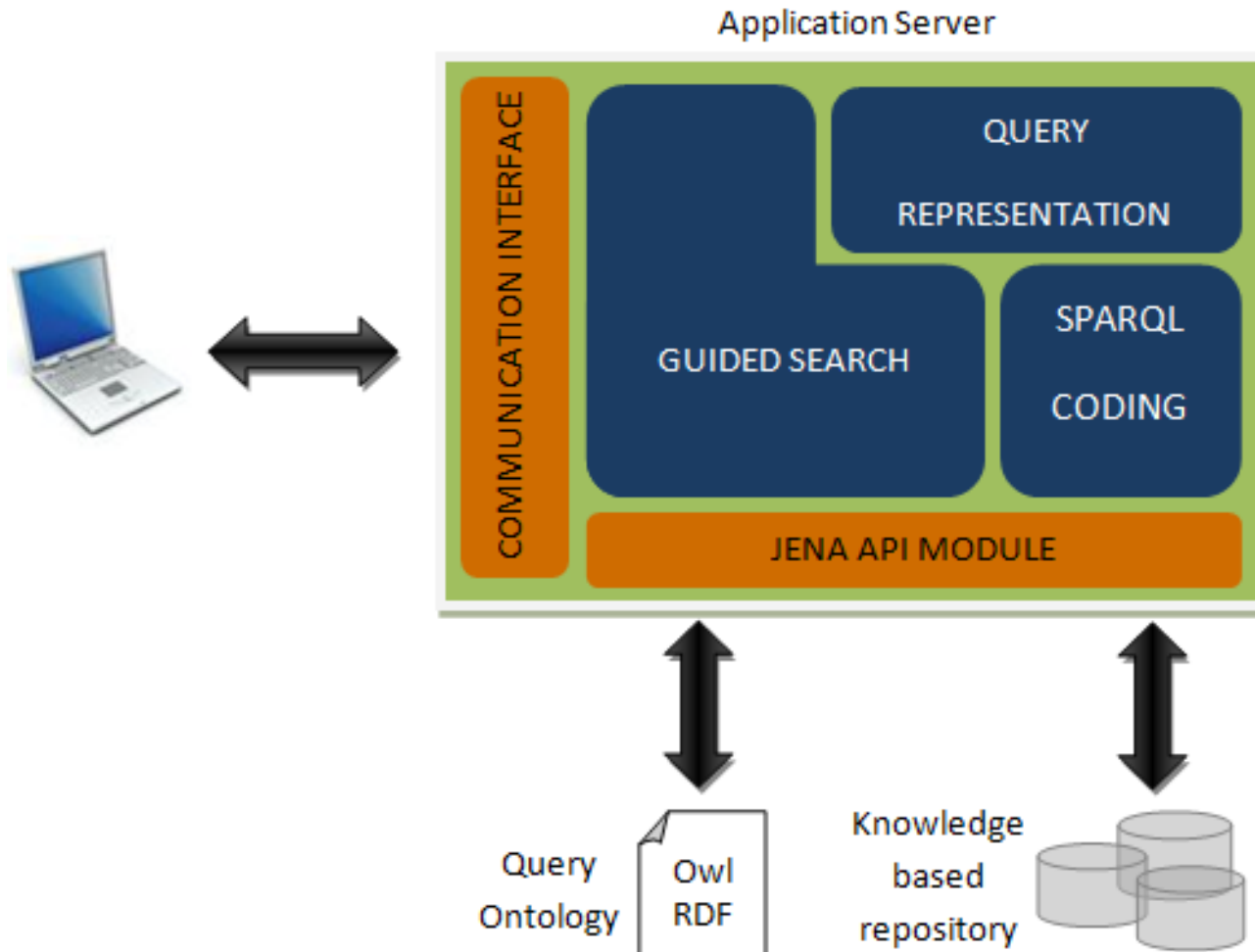
NCBI taxonomy: organisms classification

Imported ontologies: OWL punning

OGO ontology: mappings to OMIM

Implementation of the OGO system

Interfaces of the OGO system

Keyword based querying

Semantic querying

Semantic interface

Semantic interface

Sample query

Ortholog genes of the gene that causes prostate cancer on *Rattus norvegicus*?

Sample query

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```
@prefix ncbi: <http://um.es/ncbi.owl>.
@prefix ogo: <http://miuras.inf.um.es/ontologies/OGO.owl>.
SELECT
  ?Gene_0
  ?Genetic_disease_1
WHERE      {
  ?Gene_0 ogo:fromSpecies ncbi:NCBI_10116
  ?Genetic_disease_1 ogo:Name ?literal_4 .
 FILTER    (regex(?literal_4,"Prostate cancer, susceptibility to")) .
  ?Genetic_disease_1 ogo:causedBy ?Gene_2 .
  ?Cluster_of_Orthologous_genes_3 ogo:hasOrthologous ?Gene_2 .
  ?Cluster_of_Orthologous_genes_3 ogo:hasOrthologous ?Gene_0 .
}
```

Sample query

Ortholog genes of the gene that causes prostate cancer on *Rattus norvegicus*?

Genetic disease[0]	Gene[1]
104155, prostate cancer, susceptibility to, zinc finger homeobox 3	pex12, 116718
	rgd1560268, zfhx3.predicted, 307829
	rgd1563022, zfhx4.predicted, 310250
600020, neurofibrosarcoma, prostate cancer, susceptibility to, max-interacting protein 1	clec5a, 679787
	ensrnog00000026306, loc684510
	loc689617, 689617
	loc689629, 689629
	max, mgc124611, 60661
	mxi.wr, mxi1, 25701
	tgap1, 294892

Query grammar

```
Query::="SELECT" ListVar (WhereClause)?
ListVar::=Var (Var)*
WhereClause::="WHERE {" ConditionClause (ConditionClause)* "}"
ConditionClause::=[VarCondition | LiteralCondition] "."
VarCondition::=[Var | Individual] Property [Var | Individual]
LiteralCondition::=[Var | Individual] Property [Var | Individual] "."
   "FILTER (regex (" Var "," Literal "))"

Var -> This term represents a variable in the query which can be
matched to any concept or individual in the ontology.
Individual -> This term represents a concept or individual identied
by an URI in the ontology.
Property -> This term represents a relationship or property identied
by an URI in the ontology.
Literal -> This term represents any data value dened by the user.
```

Future plans

OWL reasoning for querying (OWL 2 QL?)

Pellet Integrity Constraint Validator (Pellet ICV):

  OWL as schema language for RDF (CWA)

  Check the gathered information

More bio-ontologies

Clinical archetypes for querying (ISO 13606): exchange of ortholog/disease information in a standard biomedical research setting

Conclusions

Orthologs and diseases: new hypotheses

OGO provides a resource for exploiting such combined information

Semantic query interface: "Complex" queries easily (No SPARQL syntax)

http://miuras.inf.um.es/~ogo/

Acknowledgements