W3C Semantic Web Health Care and Life Sciences Interest Group (HCLSIG)

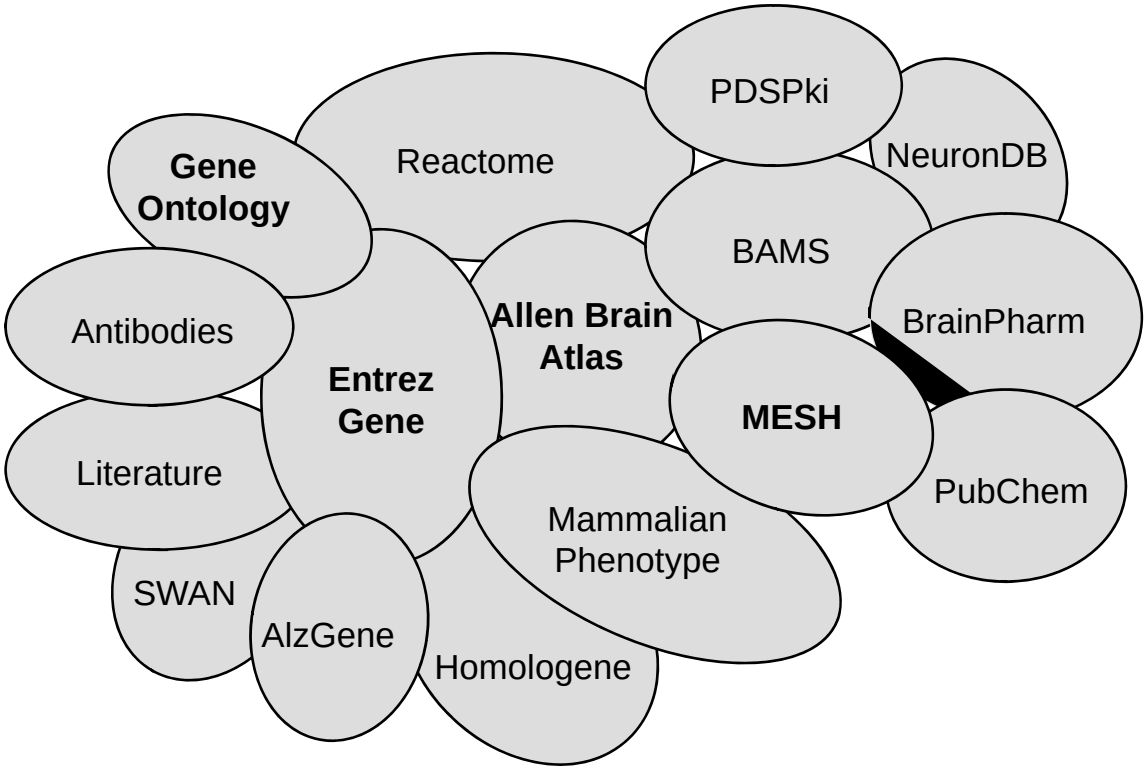
M. Scott Marshall
Eric Prud'hommeaux

HCLSIG Mission

"The mission of the Semantic Web Health Care and Life Sciences Interest Group, part of the Semantic Web Activity, is to develop, advocate, and support the use of Semantic Web technologies for biological science, translational medicine and health care. These domains stand to gain tremendous benefit by adoption of Semantic Web technologies, as they depend on the interoperability of information from many domains and processes for efficient decision support."

Scientific Questions and Sources

"Find me genes involved in signal transduction that are related to pyramidal neurons!"



Linked Data Principles

- Use URIs to name things
- Use HTTP URIs so people can look up those names
- When someone looks up a URI, provide useful RDF information
- Include RDF statements that link to other URIs so that related things can be discovered

Scientific Question in SPARQL

```
prefix go: <http://purl.org/obo/owl/GO#>
prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#>
prefix owl: <http://www.w3.org/2002/07/owl#>
prefix mesh: <http://purl.org/commons/record/mesh/>
prefix sc: <http://purl.org/science/owl/sciencecommons/>
prefix ro: <http://www.obofoundry.org/ro/ro.owl#>

select ?genename ?processname
where
{  graph <http://purl.org/commons/hcls/pubmesh>
   { ?paper ?p mesh:D017966 .
     ?article sc:identified_by_pmid ?paper.
     ?gene sc:describes_gene_or_gene_product_mentioned_by ?article.
   }
 graph <http://purl.org/commons/hcls/goa>
  { ?protein rdfs:subClassOf ?res.
    ?res owl:onProperty ro:has_function.
    ?res owl:someValuesFrom ?res2.
    ?res2 owl:onProperty ro:realized_as.
    ?res2 owl:someValuesFrom ?process.
 graph <http://purl.org/commons/hcls/20070416/classrelations>
  {{?process <http://purl.org/obo/owl/obo#part_of> go:GO_0007166}
    union
   {?process rdfs:subClassOf go:GO_0007166 }}
    ?protein rdfs:subClassOf ?parent.
    ?parent owl:equivalentClass ?res3.
    ?res3 owl:hasValue ?gene.
   }
 graph <http://purl.org/commons/hcls/gene>
  { ?gene rdfs:label ?genename }
 graph <http://purl.org/commons/hcls/20070416>
  { ?process rdfs:label ?processname}
}
```

Mesh: Pyramidal Neurons

↓

Pubmed: Journal Articles

↓

Entrez Gene: Genes

↓

GO: Signal Transduction

Inference required

W3C Semantic Web

Results

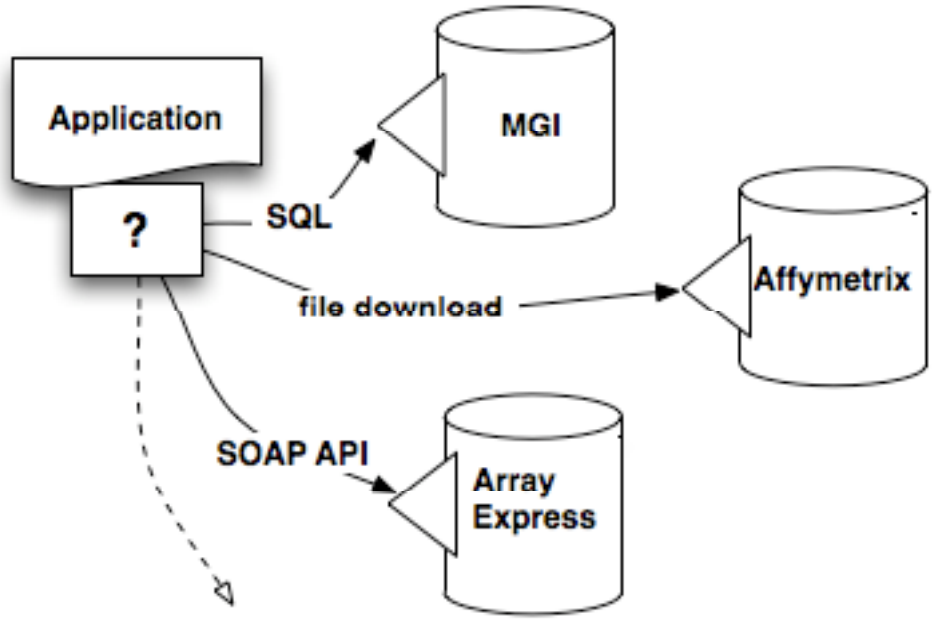
DRD1, 1812	adenylate cyclase activation
ADRB2, 154	adenylate cyclase activation
ADRB2, 154	arrestin mediated desensitization of G-protein coupled receptor protein signaling path
DRD1IP, 50632	dopamine receptor signaling pathway
DRD1, 1812	dopamine receptor, adenylate cyclase activating pathway
DRD2, 1813	dopamine receptor, adenylate cyclase inhibiting pathway
GRM7, 2917	G-protein coupled receptor protein signaling pathway
GNG3, 2785	G-protein coupled receptor protein signaling pathway
GNG12, 55970	G-protein coupled receptor protein signaling pathway
DRD2, 1813	G-protein coupled receptor protein signaling pathway
ADRB2, 154	G-protein coupled receptor protein signaling pathway
CALM3, 808	G-protein coupled receptor protein signaling pathway
HTR2A, 3356	G-protein coupled receptor protein signaling pathway
DRD1, 1812	G-protein signaling, coupled to cyclic nucleotide second messenger
SSTR5, 6755	G-protein signaling, coupled to cyclic nucleotide second messenger
MTNR1A, 4543	G-protein signaling, coupled to cyclic nucleotide second messenger
CNR2, 1269	G-protein signaling, coupled to cyclic nucleotide second messenger
HTR6, 3362	G-protein signaling, coupled to cyclic nucleotide second messenger
GRIK2, 2898	glutamate signaling pathway
GRIN1, 2902	glutamate signaling pathway
GRIN2A, 2903	glutamate signaling pathway
GRIN2B, 2904	glutamate signaling pathway
ADAM10, 102	integrin-mediated signaling pathway
GRM7, 2917	negative regulation of adenylate cyclase activity
LRP1, 4035	negative regulation of Wnt receptor signaling pathway
ADAM10, 102	Notch receptor processing
ASCL1, 429	Notch signaling pathway
HTR2A, 3356	serotonin receptor signaling pathway
ADRB2, 154	transmembrane receptor protein tyrosine kinase activation (dimerization)
PTPRG, 5793	transmembrane receptor protein tyrosine kinase signaling pathway
EPHA4, 2043	transmembrane receptor protein tyrosine kinase signaling pathway
NRTN, 4902	transmembrane receptor protein tyrosine kinase signaling pathway
CTNND1, 1500	Wnt receptor signaling pathway

Many of the genes are indeed related to Alzheimer’s Disease through gamma secretase (presenilin) activity

Demo from Browser

http://hcls.deri.ie/hcls_demo.html

Distributed Query – Before



Distributed Query – After