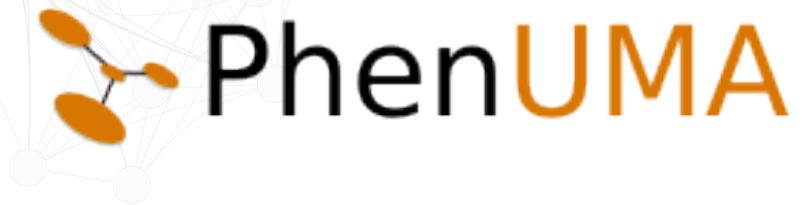




Sesión Práctica I: Introducción a



14 Junio de 2013, E.T.S.I Informática, Málaga



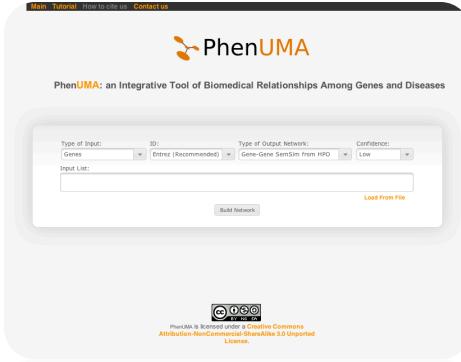
UNIVERSIDAD
DE MÁLAGA



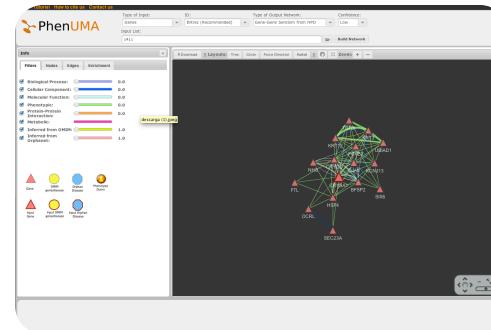
Sesión Práctica: Esquema

- **Sesión Práctica I:**
 - Entradas de Datos
 1. Opciones de Entrada
 2. Datos de Entrada
 3. Opciones de la Red de Salida
 - Knowledge Base
 - Tipos de Redes
 - Niveles de Confianza
 - Construcción de Redes
 - Funcionalidades de la Aplicación
- **Sesión Práctica II:**
 - Gestión de los Datos de Salida
 - Ejemplos y casos de uso
- **Sesión Práctica III:**
 - Ejemplos y casos de uso

Interfaz de PhenUMA



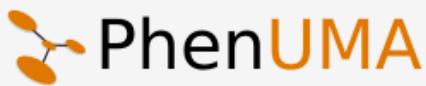
- Página Principal



- Página de Visualización

Entrada a PhenUMA

Main Tutorial How to cite us Contact us



1. Opciones de Entrada

3. Opciones de la Red de Salida

PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases

Type of Input:
Genes

ID:

Entrez (Recommended)

Type of Output Network:

Gene-Gene SemSim from HPO

Confidence:

Low

Input List:

Load From File

Build Network

2. Datos de Entrada

1. Opciones de Entrada



- Tipo de Entrada:
 - Genes
 - OMIM gene/diseases
 - Orphan Diseases
 - Phenotypes

Identificadores de Genes

- Para cada tipo de entrada debe usarse el identificador adecuado.
- Identificadores permitidos para genes:
 - Código Entrez (Recomendado)
 - Official Symbol
 - HGNC
 - MIM
 - Ensembl
 - Orphanum
- No es posible usar:
 - Full Name: crystallin, beta A1
 - Sinónimos: CRYB1, CTRCT10
 - Proteína: Beta-crystallin A3

Identificadores de Genes



¿Cómo obtener los identificadores?

NCBI:

<http://www.ncbi.nlm.nih.gov/>

Ensembl

<http://www.ensembl.org/>

HGNC

<http://www.genenames.org/>

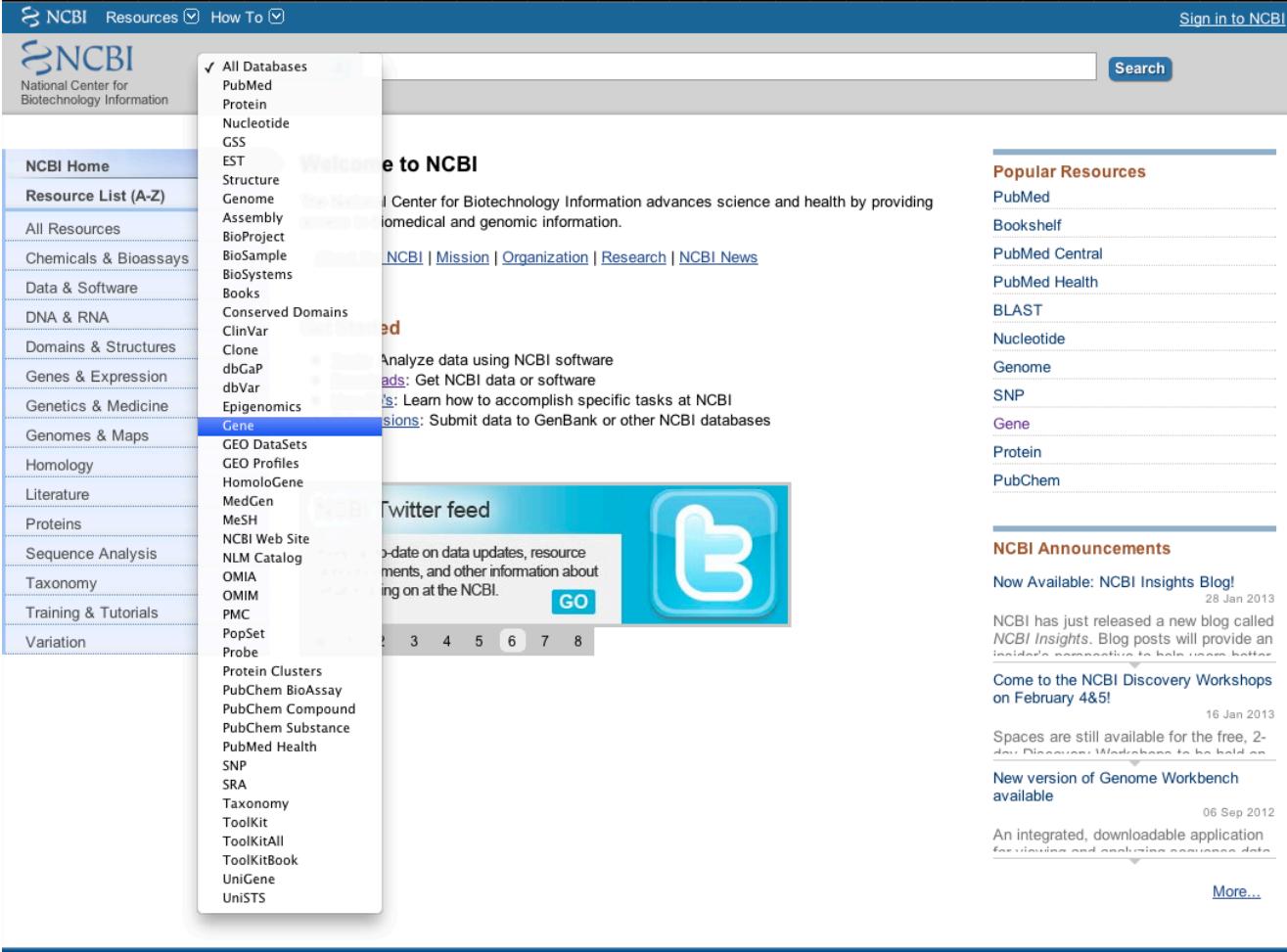
MIM

<http://www.omim.org/>

Orphanet

<http://www.orpha.net/>

Identificadores de Genes: NCBI



The screenshot shows the NCBI homepage with a sidebar menu on the left. The 'Gene' option is highlighted in blue, indicating it is selected. The main content area features a search bar at the top right, followed by a brief introduction about NCBI's mission to advance science and health through biomedical and genomic information. Below this is a 'Twitter feed' section with a blue header and a large Twitter logo. To the right of the main content are two columns: 'Popular Resources' and 'NCBI Announcements'. The 'Popular Resources' column lists links to PubMed, Bookshelf, PubMed Central, PubMed Health, BLAST, Nucleotide, Genome, SNP, Gene, Protein, and PubChem. The 'NCBI Announcements' column lists three recent posts: 'Now Available: NCBI Insights Blog!', 'Come to the NCBI Discovery Workshops on February 4&5!', and 'New version of Genome Workbench available'.

NCBI Resources How To

NCBI National Center for Biotechnology Information

NCBI Home Resource List (A-Z)

All Resources Chemicals & Bioassays Data & Software DNA & RNA Domains & Structures Genes & Expression Genetics & Medicine Genomes & Maps Homology Literature Proteins Sequence Analysis Taxonomy Training & Tutorials Variation

All Databases

- PubMed
- Protein
- Nucleotide
- GSS
- EST
- Structure
- Genome
- Assembly
- BioProject
- BioSample
- BioSystems
- Books
- Conserved Domains
- ClinVar
- Clone
- dbGaP
- dbVar
- Epigenomics
- Gene
- GEO DataSets
- GEO Profiles
- HomoloGene
- MedGen
- MeSH
- NCBI Web Site
- NLM Catalog
- OMIA
- OMIM
- PMC
- PopSet
- Probe
- Protein Clusters
- PubChem BioAssay
- PubChem Compound
- PubChem Substance
- PubMed Health
- SNP
- SRA
- Taxonomy
- Toolkit
- ToolkitAll
- ToolkitBook
- UniGene
- UniSTS

Search

Welcome to NCBI

NCBI advances science and health by providing biomedical and genomic information.

NCBI | Mission | Organization | Research | NCBI News

Analyze data using NCBI software

ads: Get NCBI data or software

ts: Learn how to accomplish specific tasks at NCBI

sions: Submit data to GenBank or other NCBI databases

Twitter feed

Get up-to-date on data updates, resource announcements, and other information about what's happening on at the NCBI.

GO

1 2 3 4 5 6 7 8

Popular Resources

PubMed

Bookshelf

PubMed Central

PubMed Health

BLAST

Nucleotide

Genome

SNP

Gene

Protein

PubChem

NCBI Announcements

Now Available: NCBI Insights Blog! 28 Jan 2013

NCBI has just released a new blog called *NCBI Insights*. Blog posts will provide an integrated commentary to help users better

Come to the NCBI Discovery Workshops on February 4&5! 16 Jan 2013

Spaces are still available for the free, 2-day *Discovery Workshops* to be held on

New version of Genome Workbench available 06 Sep 2012

An integrated, downloadable application for viewing and analyzing sequence data

More...

Entrada a PhenUMA → 1. Opciones de la Entrada → Identificadores de Genes

Identificadores de Genes: NCBI

The screenshot shows the NCBI Gene search results for the gene CRYBA1. The search bar at the top has 'Gene' selected. The main result is for 'CRYBA1 crystallin, beta A1 [Homo sapiens (human)]'. The 'Entrez/Gene ID' is listed as 1411, updated on 9-Jun-2013. The 'Official Symbol' is CRYBA1. The 'Official Full Name' is crystallin, beta A1. The 'Primary source' is HGNC:2394. The 'See related' section lists Ensembl:ENSG00000108255, HPRD:00429, MIM:123610, Vega:OTTHUMG00000132729. The 'Gene type' is protein coding, 'RefSeq status' is REVIEWED, and the 'Organism' is Homo sapiens. The 'Lineage' includes Eukaryota, Metazoa, Chordata, Craniata, Vertebrata, Euteleostomi, Mammalia, Eutheria, Euarchontoglires, Primates, Haplorrhini, Catarrhini, Hominidae, Homo. The 'Also known as' section lists CRYB1, CTRC10. The 'Summary' section contains a detailed description of crystallins, mentioning their taxon-specific nature, transparency, and refractive index. It also describes the different families of crystallins (alpha, beta, gamma) and their subfamilies (acidic and basic). The gene encodes two proteins (crystallin, beta A3 and crystallin, beta A1) from a single mRNA. Deletion of exons 3 and 4 causes the autosomal dominant disease 'zonular cataract with sutural opacities'. The right sidebar provides links to Table of contents, Summary, Genomic context, Genomic regions, transcripts, and products, Bibliography, Phenotypes, Interactions, General gene information, General protein information, Reference sequences, Related sequences, Additional links, and Related Information.

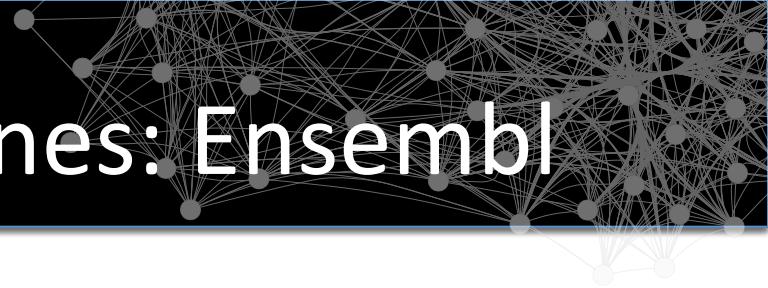
Entrez/Gene ID

Official Symbol

Más Identificadores

Identificadores de Genes: Ensembl

<http://www.ensembl.org/>



The screenshot shows the Ensembl homepage. At the top, there's a search bar with "Human" selected, looking for "Beta-crystallin A3". Below the search bar, there are sections for "Browse a Genome", "Popular genomes" (Human, Mouse, Zebrafish), and "New to Ensembl?" with links to learn how to use Ensembl, add custom tracks, upload data, search for DNA or protein sequences, fetch data, download databases via FTP, and mine Ensembl with BioMart. To the right, there's a sidebar for "What's New in Release 71 (April 2013)" which includes links to new expression views, new assemblies for Chicken and C.elegans, and a scrollable image integration. There are also links to full details of the release, more release news, and latest blog posts. A "Did you know..." box at the bottom right explains how to adjust the width of the page.

Search: Human for Beta-crystallin A3 Go
e.g. BRCA2 or rat X:100000..200000 or coronary heart disease

Browse a Genome
The Ensembl project produces genome databases for vertebrates and other eukaryotic species, and makes this information freely available online.

Popular genomes

New to Ensembl?

- Learn how to use Ensembl
- Add custom tracks
- Upload and analyse your data
- Search for a DNA or protein sequence
- Fetch only the data you want
- Download our databases via FTP
- Mine Ensembl with BioMart

Still got questions? Try our [FAQs](#) or [glossary](#)

What's New in Release 71 (April 2013)

- New Expression view listing RNAseq data
- New assemblies for Chicken (Galgal4) and C.elegans (WBcel235)
- Scrollable image has been integrated into Region in Detail

[Full details of this release](#)
[More release news on our blog →](#)

Latest blog posts

- 06 Jun 2013: [Ensembl archive sites offline](#)
- 04 Jun 2013: [Ensembl is the place!](#)
- 12 May 2013: [Retirement of archive 58](#)

[Go to Ensembl blog →](#)

Did you know...?
Adjust the width of your display. Click [configure this page](#) from most [views](#), then the [Display options](#) menu.

Identificadores de Genes: Ensembl

Resultado de la búsqueda de “Beta-crystallin A3”

http://www.ensembl.org/Homo_sapiens/Search/Results?species=Homo_sapiens;idx=;q=Beta-crystallin%20A3



The screenshot shows the Ensembl search results for "Beta-crystallin A3" in the Human (GRCh37) database. The results are summarized in two tables: one by Feature type and one by Species.

Results Summary

Your search of Human with 'Beta-crystallin A3' returned the following results:

By Feature type	
Total	2
Gene	1
Human (1)	
Transcript	1
Human (1)	

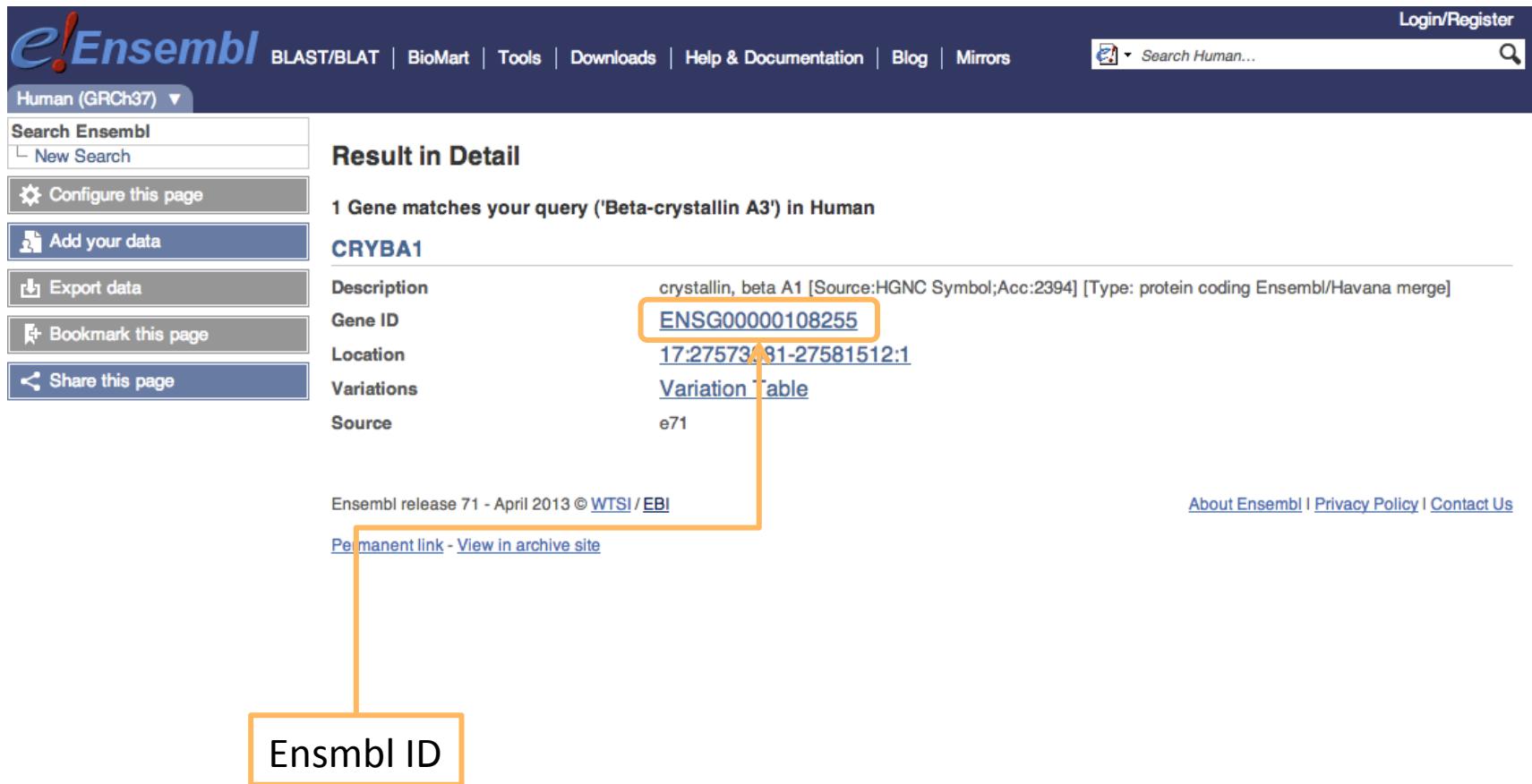
By Species	
Total	2
Human	2
Gene (1)	
Transcript (1)	

Ensembl release 71 - April 2013 © WTSI / EBI

[About Ensembl](#) | [Privacy Policy](#) | [Contact Us](#)

[Permanent link](#) - [View in archive site](#)

Identificadores de Genes: Ensembl



The screenshot shows the Ensembl gene detail page for the gene CRYBA1. The page has a dark blue header with the Ensembl logo and navigation links like BLAST/BLAT, BioMart, Tools, Downloads, Help & Documentation, Blog, and Mirrors. A search bar is also present. The main content area is titled "Result in Detail" and shows that 1 gene matches the query "Beta-crystallin A3" in Human. The gene is identified as CRYBA1, which is described as crystallin, beta A1. Its Ensembl ID is ENSG00000108255, located on chromosome 17 at position 27573181-27581512. A Variation Table link is provided. The page footer includes links to Ensembl release 71 (April 2013), WTSI/EBI, About Ensembl, Privacy Policy, and Contact Us. An orange box highlights the Ensembl ID (ENSG00000108255) and the permanent link (Permanent link - View in archive site). Another orange box highlights the "Ensembl ID" label below the page.

Human (GRCh37) ▾

Search Ensembl
New Search

Configure this page

Add your data

Export data

Bookmark this page

Share this page

Result in Detail

1 Gene matches your query ('Beta-crystallin A3') in Human

CRYBA1

Description: crystallin, beta A1 [Source:HGNC Symbol;Acc:2394] [Type: protein coding Ensembl/Havana merge]

Gene ID: [ENSG00000108255](#)

Location: [17:27573181-27581512:1](#)

Variations: [Variation Table](#)

Source: e71

Ensembl release 71 - April 2013 © [WTSI / EBI](#)

[About Ensembl](#) | [Privacy Policy](#) | [Contact Us](#)

Permanent link - [View in archive site](#)

Ensembl ID

Identificadores de Genes: HGNC

<http://www.genenames.org/>

The screenshot shows the HGNC (HUGO Gene Nomenclature Committee) website. At the top, there is a navigation bar with links: Home, Search Genes, Downloads, Gene Families, HCOP, Useful Links, About, Contact Us, and Request Symbol. Below the navigation bar, there is a search bar labeled "Search Genes" with a magnifying glass icon. To the left of the search bar, there is a small image of a karyogram showing human chromosomes. To the right of the search bar, there is a text block that reads: "The HUGO Gene Nomenclature Committee (HGNC) has assigned unique gene symbols and names to over 37,000 human loci, of which around 19,000 are protein coding. [genenames.org](#) is a curated online repository of HGNC-approved gene nomenclature and associated resources including links to genomic, proteomic and phenotypic information, as well as dedicated gene family pages." At the bottom left, there is a link "Browse approved symbols by chromosome". On the right side, there is a "Quick Gene Search" form with a search input field containing "crystallin|beta A1" and a search button.

Identificadores de Genes: HGNC

<http://www.genenames.org/>



The HGNC website interface is shown. At the top, there is a navigation bar with links to Home, Search Genes, Downloads, Gene Families, HGCP, Useful Links, About, Contact Us, and Request Symbol. Below the navigation bar is a search bar labeled "Search Genes" with a magnifying glass icon. The main content area is titled "Quick Gene Search". It features a search input field containing "crystallin beta A1" and a search button. Below the search form, there are links for "First Page", "Prev Page", page numbers 1, 2, 3, 4, ..., 69, and "Next Page". The text "Total hits: 3441" is displayed. A table follows, showing gene information with columns for Approved Symbol, Approved Name, Location, and Best Match. The table rows are:

Approved Symbol	Approved Name	Location	Best Match
CRYBA1	crystallin, beta A1	17q11.2-q12	Approved Name: crystallin, beta A1
CRYBG3	beta-gamma crystallin domain containing 3	3q11.2	Approved Name: beta-gamma crystallin domain containing 3
CRYBA2	crystallin, beta A2	2q35	Approved Name: crystallin, beta A2
CRYBA4	crystallin, beta A4	22q12.1	Approved Name: crystallin, beta A4
CRYBB1	crystallin, beta B1	22q12.1	Approved Name: crystallin, beta B1
CRYBB2	crystallin, beta B2	22q11.23	Approved Name: crystallin, beta B2
CRYBB2P1	crystallin, beta B2 pseudogene 1	22q11.2-q12.1	Approved Name: crystallin, beta B2 pseudogene 1

Identificadores de Genes: HGNC

<http://www.genenames.org/>

The screenshot shows the HGNC Gene Symbol Report for the gene CRYBA1. The page has a dark blue header with the HGNC logo and a search bar. Below the header, there's a navigation menu with links to Home, Search Genes, Downloads, Gene Families, HGCP, Useful Links, About, Contact Us, and Request Symbol. The main content area is titled "Gene Symbol Report" and displays the gene symbol "CRYBA1". To the left of the main content, three boxes are overlaid: "Symbol" points to the gene symbol, "HGNC ID" points to the HGNC ID (2394), and "Más Identificadores" points to the "GENE RESOURCES" section which lists various databases and their links.

Gene Symbol Report

CRYBA1

Approved Symbol: CRYBA1
Approved Name: crystallin, beta A1
HGNC ID: 2394
Previous Symbols & Names: CRYB1
Synonyms: "eye lens structural protein"
Locus Type: gene with protein product
Chromosomal Location: 17q11.2-q12

HOMOLOGS: MGI:88518 C Mouse Symbol: Cryba1
RGD:2415 D Rat Symbol: Cryba1
HGCP D
TreeFam D

NUCLEOTIDE SEQUENCES: RefSeq:NM_005208 C
CCDS:CCDS11249.1 C
Vena:OTTHUMG00000132729 C

GENE RESOURCES: Entrez Gene:1411 C
Ensembl:ENSG00000108255 C
UCSC:uc002hdw.3 D
Vega:OTTHUMG00000132729 C
NCBI Sequence Viewer
Ensembl Genome Browser
UCSC Genome Browser
Vega Genome Browser

PROTEIN RESOURCES: UniProtKB:P05813 D
InterPro D

CLINICAL RESOURCES: OMIM D
GeneTests D
Orphanet D
DECIPHER D
COSMIC D
LSDB:LOVD - Leiden Open Variation Database C
Genetic Testing Registry C

Entrada a PhenUMA → 1. Opciones de la Entrada → Identificadores de Genes

Identificadores de Genes: MIM

<http://www.omim.org/>

Home | About | Statistics ▾ | Downloads/API ▾ | Help ▾ | External Links | Terms of Use ▾ | Contact Us

 Seleccionar idioma ▾

Mirror sites: us-east.omim.org, europe.omim.org

OMIM®

Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and Genetic Disorders
Updated 10 June 2013

beta crystallin a1

Search

Sample Searches
OMIM Tutorial

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map , Search History





Entrada a PhenUMA → 1. Opciones de la Entrada → Identificadores de Genes

Identificadores de Genes: MIM

<http://www.omim.org/>

Home | About | Statistics | Downloads/API | Help | External Links | Terms of Use | Contact Us | [Seleccionar idioma](#)

beta crystallin a1 [Search](#) Sort by: Relevance Date updated
Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Toggle: search terms highlighted
Search History: View, Clear Retrieve corresponding: [gene map](#) [clinical synopses](#)

Search: 'beta crystallin a1'
Results: 1 - 10 of 4,454 | [Show top 100](#) | [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [Next](#) [Last](#)

1 :	* 123610. CRYSTALLIN, BETA-A1; CRYBA1 Cytogenetic location: 17q11.2 , Genomic coordinates (GRCh37): 17:27,573,874 - 27,581,511 Matching terms: a1, crystallin, beta	Links
2 :	* 123620. CRYSTALLIN, BETA-B2; CRYBB2 CRYBB2P1, INCLUDED Cytogenetic location: 22q11.23 , Genomic coordinates (GRCh37): 22:25,615,611 - 25,627,835 Matching terms: a1, crystallin, beta	Links
3 :	# 600881. CATARACT 10, MULTIPLE TYPES; CTRCT10 Cytogenetic location: 17q11.2 Matching terms: a1, crystallin, beta	Links
4 :	* 123630. CRYSTALLIN, BETA-B3; CRYBB3 Cytogenetic location: 22q11.23 , Genomic coordinates (GRCh37): 22:25,595,824 - 25,603,323 Matching terms: a1, crystallin, beta	Links
5 :	* 600836. CRYSTALLIN, BETA-A2; CRYBA2 Cytogenetic location: 2q35 , Genomic coordinates (GRCh37): 2:219,854,911 - 219,858,126 Matching terms: a1, crystallin, beta	Links

Identificadores de Genes: MIM

<http://www.omim.org/>

OMIM ID

Symbol

beta crystallin a1

Search Sort by: Relevance Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Toggle: search terms highlighted, changes highlighted

Search History: View, Clear

*123610

CRYSTALLIN, BETA-A1; CRYBA1

Alternative titles; symbols

CRYSTALLIN, BETA-1; CRYB1

CRYSTALLIN, BETA-A1/A3

HGNC Approved Gene Symbol: CRYBA1

Cytogenetic location: 17q11.2 Genomic coordinates (GRCh37): 17:27,573,874 - 27,581,511 (from NCB)

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
17q11.2	Cataract 10, multiple types	600881

Table of Contents - *123610

External Links:

- ▶ Genome
- ▶ DNA
- ▶ Protein
- ▶ Gene Info
- ▶ Clinical Resources
- ▶ Variation
- ▶ Animal Models
- ▶ Cellular Pathways

Identificadores de Genes: Orphanum

<http://www.orpha.net/>

Languages: FR EN ES | DE IT PT

Homepage
About Orphanet
Help
Contact us

Rare diseases are rare, but rare disease patients are numerous

Access our Services

Inventory, classification and encyclopediea of rare diseases, with genes involved

Inventory of ongoing research projects, clinical trials, registries and blobanks

Directory of patient organisations

Assistance-to-diagnosis tool

Emergency guidelines

Inventory of orphan drugs

Directory of medical laboratories providing diagnostic tests

Directory of expert centres

Newsletter

Collection of thematic reports: Orphanet Reports Series

Read Orphanet reports

List of rare diseases

Prevalence of Rare Diseases

Disease registries in Europe

European research projects & clinical networks

Lists of medicinal products

Orphanet Activity Reports

Satisfaction Surveys

Contribute to Orphanet

Register your activity

Sponsor Orphanet

Download Orphanet data

Orphadata

Newsletter

Read the last newsletter

Read previous issues

Sign up to receive the newsletter

Other documents

Council Recommendation on an action in the field of rare diseases

State of Art of rare diseases

Other rare diseases websites

Rare Diseases - European Commission

EUCERD

European Medicines Agency

IRDIRC

Office of rare diseases research (US)

Events

See all

JULY 1 Myasthenia 2013
1 - 2 July 2013, Paris, France

Búsquedas de Genes y Enfermedades

Entrada a PhenUMA → 1. Opciones de la Entrada → Identificadores de Genes

Identificadores de Genes: Orphanum

<http://www.orpha.net/>

The screenshot shows the Orphanet homepage with several orange boxes highlighting specific features:

- An orange box surrounds the "Genes" link in the top navigation bar.
- An orange box surrounds the "Simple Search" form, which contains a field for entering a gene name or symbol ("beta crystallin a1") and a dropdown menu for selecting the type of identifier (radio buttons for "Gene name or symbol", "MIM number (Gene)", "Disease name", and "MIM number (disease)").
- An orange box surrounds the search results for the gene "CRYBA1 - Crystallin, beta A1", displaying various identifiers and their corresponding values.

Identifier	Value
Orpha number	ORPHA120836
OMIM	123610 [↗]
HGNC	2394 [↗]
UniProtKB	P05813 [↗]
Genatlas	CRYBA1 [↗]
Ensembl	ENSG00000108255 [↗]
IUPHAR-DB	-
Reactome	-

Indicar tipo de búsqueda: Gen

Formulario de búsqueda: Indicar tipo de identificador

Salida: Código ORPHA, OMIM, HGNC, Ensembl....

Identificadores de Enfermedades: OMIM

- Enfermedades OMIM
 - <http://www.omim.org/>
 - Buscar por nombre de enfermedad:
 - Ej: Friedreich ataxia

Home | About | Statistics ▾ | Downloads/API ▾ | Help ▾ | External Links | Terms of Use ▾ | Contact Us

friedreich ataxia Sort by: Relevance Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Toggle: search terms highlighted
Search History: View, Clear

#229300

FRIEDREICH ATAXIA 1; FRDA

Alternative titles; symbols

FRDA1
FA

Other entities represented in this entry:

FRIEDREICH ATAXIA WITH RETAINED REFLEXES, INCLUDED; FARR, INCLUDED

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
9q21.11	Friedreich ataxia with retained reflexes	229300	FXN	606829
9q21.11	Friedreich ataxia	229300	FXN	606829

Identificadores de Enfermedades: Orphanet

- Enfermedades Raras

The screenshot shows the Orphanet homepage with a navigation bar at the top. Below the header, there's a banner with the text "Rare diseases are rare, but rare disease patients are numerous". The main content area features a search interface. In the "SIMPLE SEARCH" section, the term "Friedreich ataxia" is entered into a field, and the "Disease name" radio button is selected. An "OK" button is visible next to the search results. The results are presented in a table format under the heading "OTHER SEARCH OPTION(S)".

Orpha number	:	ORPHA95	ICD-10	:	G11.1
Synonym(s)	:	-	OMIM	:	229300 601992
Prevalence	:	1-9 / 100 000	UMLS	:	C0016719
Inheritance	:	Autosomal recessive	MeSH	:	D005621
Age of onset	:	Childhood	MedDRA	:	10017374
			SNOMED CT	:	10394003

Entrada a PhenUMA → 1. Opciones de la Entrada → Identificadores de Genes

Fenotipos

- PhenExplorer
 - <http://compbio.charite.de/phenexplorer/>

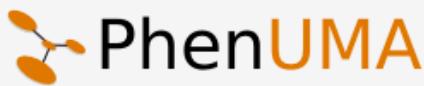
The screenshot shows a web-based search interface for phenotypes. At the top, there are four tabs: "by features." (which is selected), "by disease.", "by ontology.", and "by genes.". Below the tabs is a search bar labeled "Enter feature" with a "search." button and a "reset." button. The main area displays a table of phenotype entries:

HPO-ID	Feature
HP:0010704	1-2 finger syndactyly
HP:0005767	1-2 toe complete cutaneous syndactyly
HP:0010711	1-2 toe syndactyly
HP:0010706	1-3 finger syndactyly
HP:0001459	1-3 toe syndactyly
HP:0010707	1-4 finger syndactyly
HP:0010712	1-4 toe syndactyly
HP:0006088	1-5 finger complete cutaneous syndactyly
HP:0010708	1-5 finger syndactyly
HP:0010713	1-5 toe syndactyly
HP:0000878	11 pairs of ribs
HP:0001233	2-3 finger syndactyly

At the bottom left, there are navigation icons for first, previous, next, last, and search. The page number "Page 1" and "of 842" are displayed. On the right, it says "Displaying features 1 - 12 of 10100".

Entrada a PhenUMA

Main Tutorial How to cite us Contact us



1. Opciones de Entrada

3. Opciones de la Red de Salida

PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases

Type of Input:
Genes

ID:

Entrez (Recommended)

Type of Output Network:

Gene-Gene SemSim from HPO

Confidence:

Low

Input List:

Load From File

Build Network

2. Datos de Entrada

2. Datos de Entrada

- Separada por comas:
 - Ejemplo1
 - SLC7A7, SLC7A9, FXN, GBA, SCARB2, SLC3A1, SLC16A2

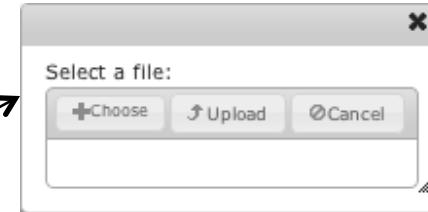
The screenshot shows the PhenUMA web application interface. At the top, there is a navigation bar with links: Main, Tutorial, How to cite us, and Contact us. Below the navigation bar is the PhenUMA logo, which consists of a stylized orange and yellow molecular structure icon followed by the text "PhenUMA". A subtitle below the logo reads "PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases". The main content area contains a form for input parameters. The "Type of Input:" dropdown is set to "Genes". The "ID:" dropdown is set to "GeneSymbol (Official Sy)". The "Type of Output Network:" dropdown is set to "Gene-Gene SemSim from HPO". The "Confidence:" dropdown is set to "Low". In the "Input List:" text input field, the genes SLC7A7, SLC7A9, FXN, GBA, SCARB2, SLC3A1, and SLC16A2 are listed. There are two buttons at the bottom of the form: "Load From File" and "Build Network".

2. Datos de Entrada

- Cargar entrada desde fichero de texto

The screenshot shows the PhenUMA web application's input configuration panel. It includes fields for 'Type of Input' (set to 'Genes'), 'ID' (set to 'GeneSymbol (Official Symbol)'), 'Type of Output Network' (set to 'Gene-Gene SemSim from HPO'), and 'Confidence' (set to 'Low'). Below these are dropdown menus for 'Input List' and 'Build Network'. A prominent blue button labeled 'Load From File' is highlighted with a yellow border. At the bottom left is a Creative Commons Attribution-NonCommercial-ShareAlike 3.0 Unported License logo, and at the bottom right is a note about the license.

1. Presionar "Load From File"



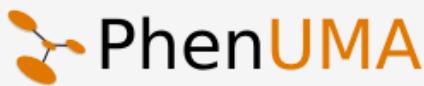
2. En la ventana emergente pulsar "Choose" para seleccionar el archivo



3. "Upload" para cargar el archivo seleccionado y "Cancel" para eliminar el archivo seleccionado

Entrada a PhenUMA

Main Tutorial How to cite us Contact us



1. Opciones de Entrada

3. Opciones de la Red de Salida

PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases

Type of Input:
Genes

ID:

Entrez (Recommended)

Type of Output Network:

Gene-Gene SemSim from HPO

Confidence:

Low

Input List:

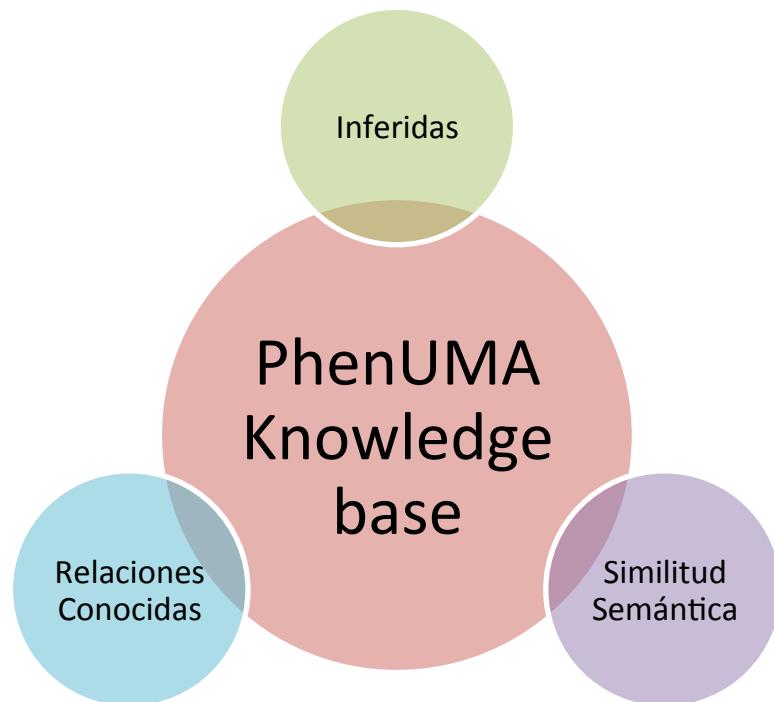
Load From File

Build Network

2. Datos de Entrada

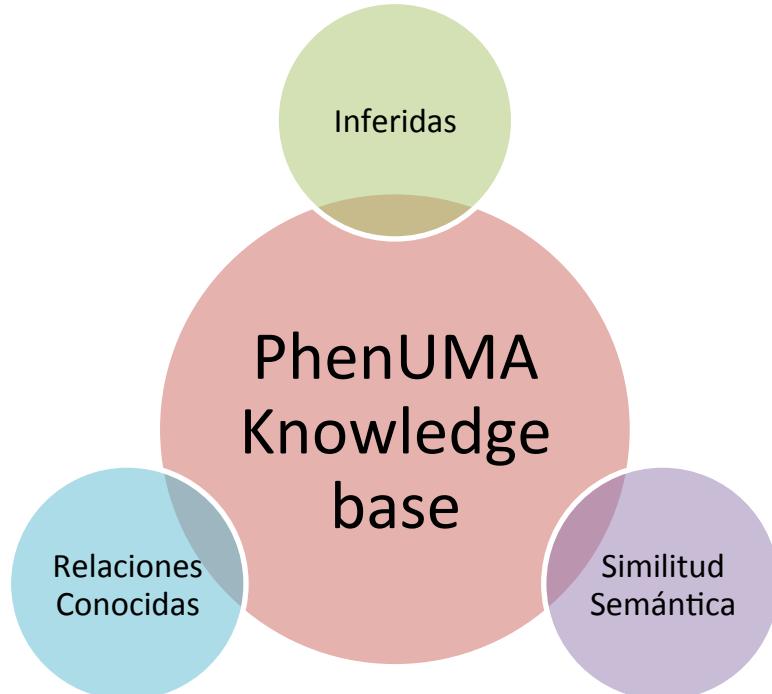
3. Opciones de Salida: Knowledge Base

- Las relaciones entre genes y enfermedades se clasifican en tres grupos:



Knowledge Base

Gene-Gene Inferred from OMIM
Gene-Gene Inferred from Orphanet

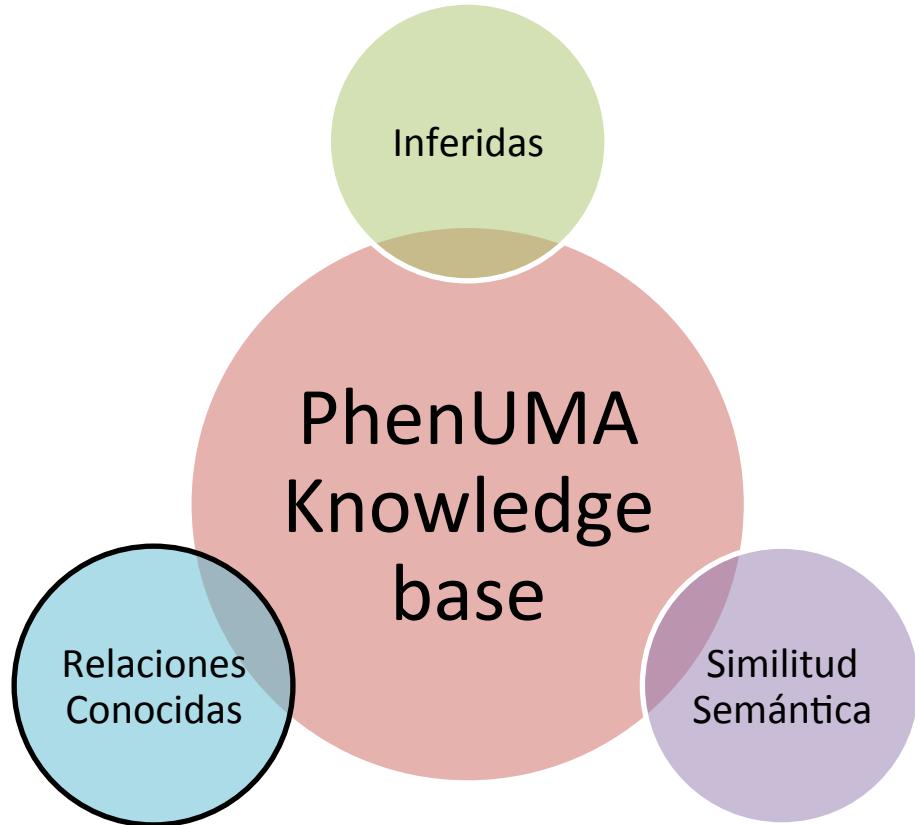


Gene-Gene Known Protein-Protein Interaction from STRING
Gene-Gene Known Metabolic Interactions (Veeramani et al.)

Gene-OMIM Known OMIM Relationships
Gene-ORPHA Known from Orphanet

Gene-Gene SimSem from HPO
Gene-Gene SimSem from GO (Biological Process)
Gene-Gene SimSem from GO (Cellular Component)
Gene-Gene SimSem from GO (Molecular Function)

3. Opciones de Salida: Knowledge Base



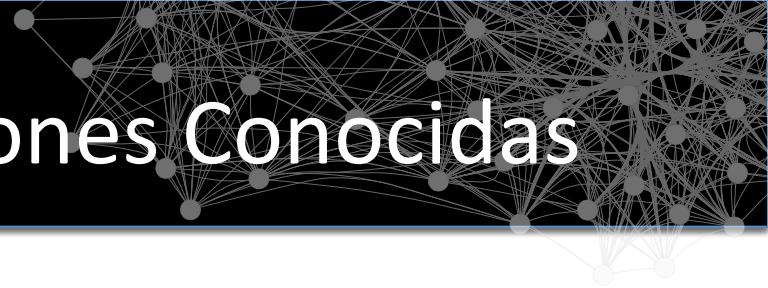
Knowledge Base : Relaciones Conocidas

- STRING:
 - Base de datos que incluye información de proteínas que interaccionan físicamente.
 - Humanos (*Homo sapiens*)
 - 96000 interacciones aproximadamente
- Metabólicas (Veeramani et al.)
 - Recon 1
 - Futuro : Recon2



<http://string-db.org/>

Knowledge Base : Relaciones Conocidas



- Relaciones gen-OMIM y gen-Orphan.
- Relaciones moleculares entre variaciones genéticas y expresión de fenotipos.



Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and Genetic Disorders
Updated 7 June 2013

Search

Sample Searches
OMIM Tutorial

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map

<http://www.omim.org/>



Knowledge Base : Relaciones Conocidas

#271980

ICD+

SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY; SSADHD

Alternative titles; symbols

SSADH DEFICIENCY
4-HYDROXYBUTYRIC ACIDURIA
GABA METABOLIC DEFECT
GAMMA-HYDROXYBUTYRIC ACIDURIA

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
6p22.3	Succinic semialdehyde dehydrogenase deficiency	271980	ALDH5A1	610045

Clinical Synopsis

TEXT

A number sign (#) is used with this entry because succinic semialdehyde dehydrogenase (SSADH) deficiency can be caused by homozygous mutation in the ALDH5A1 gene ([610045](#)) on chromosome 6p22.

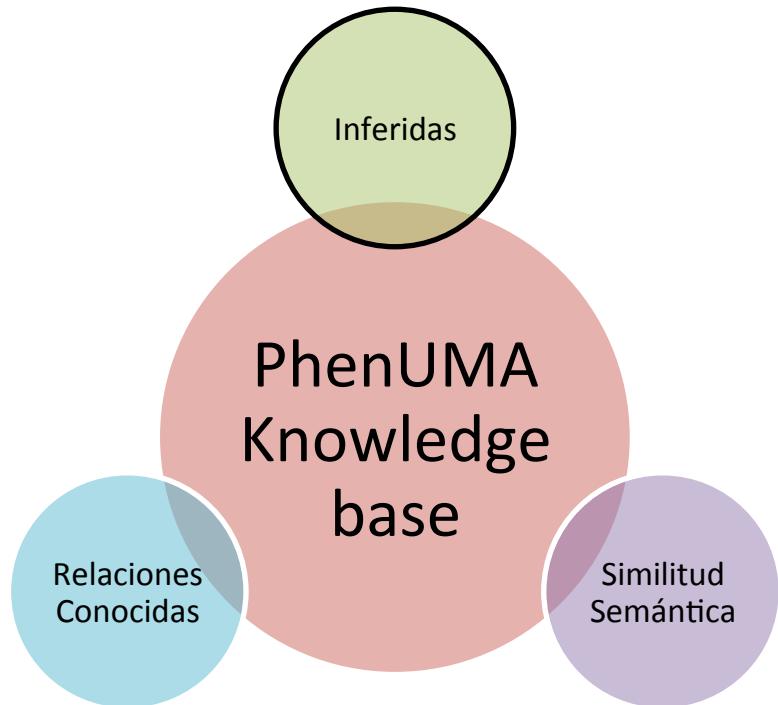
Description

Succinic semialdehyde dehydrogenase deficiency (SSADHD) is a rare autosomal recessive neurologic disorder in which an enzyme defect in the GABA degradation pathway causes a consecutive elevation of gamma-hydroxybutyric acid (GHB) and GABA. The clinical features include developmental delay, hypotonia, mental retardation, ataxia, seizures, hyperkinetic behavior, aggression, and sleep disturbances (summary by [Reis et al., 2012](#)).

<http://www.omim.org/entry/271980?search=271980&highlight=271980>

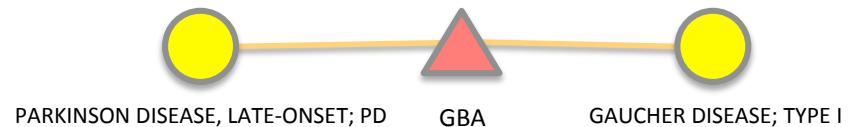
3. Opciones de Salida: Knowledge Base

- Relaciones Inferidas: a partir de las conocidas



Knowledge Base : Relaciones Inferidas

- A partir de las relaciones conocidas de OMIM y Orphanet



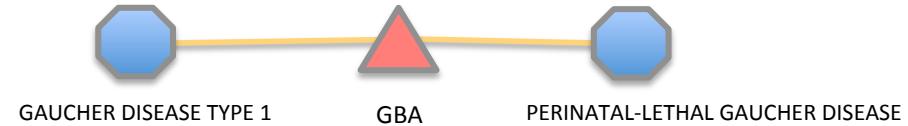
Gene-Gene Inferred from OMIM



OMIM-OMIM Inferred from Gene

Knowledge Base : Relaciones Inferidas

- A partir de las relaciones conocidas de OMIM y Orphanet

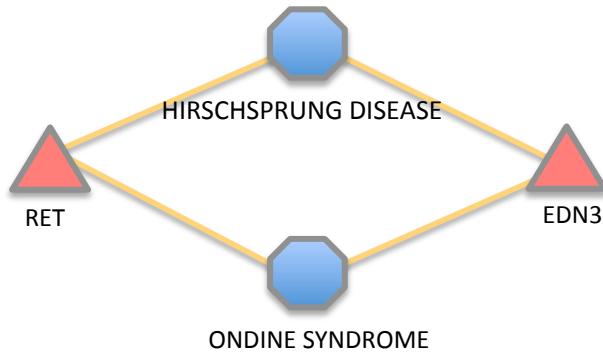


Gene-Gene Inferred from Orphanet

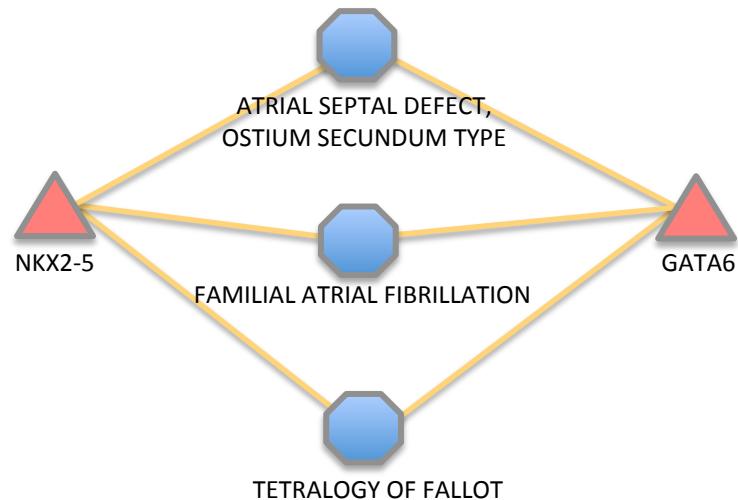
Orpha-Orpha Inferred from Gene

Knowledge Base : Relaciones Inferidas

- Relaciones múltiples.
- Valor de la relación determinado por el número de enfermedades compartidas

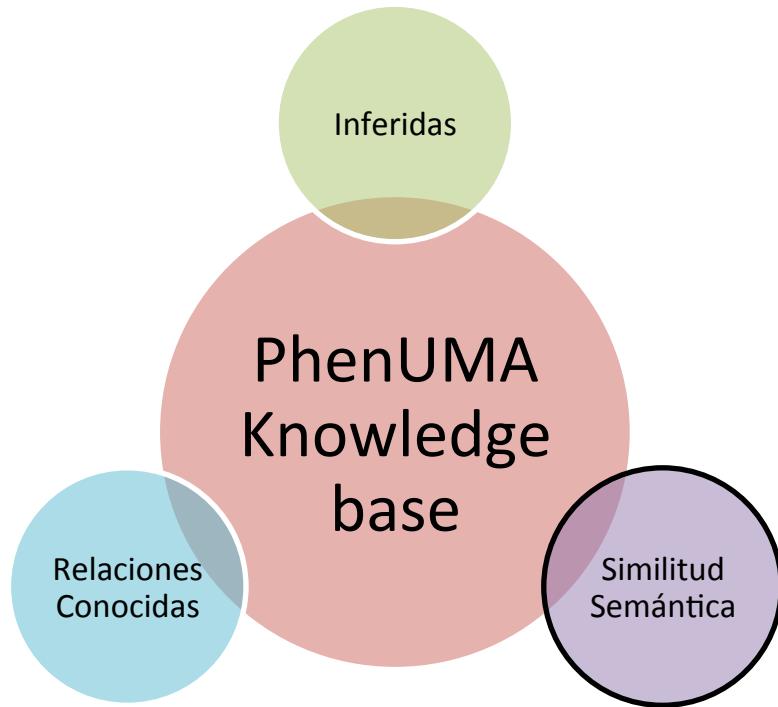


Gene-Gene Inferred from Orphanet



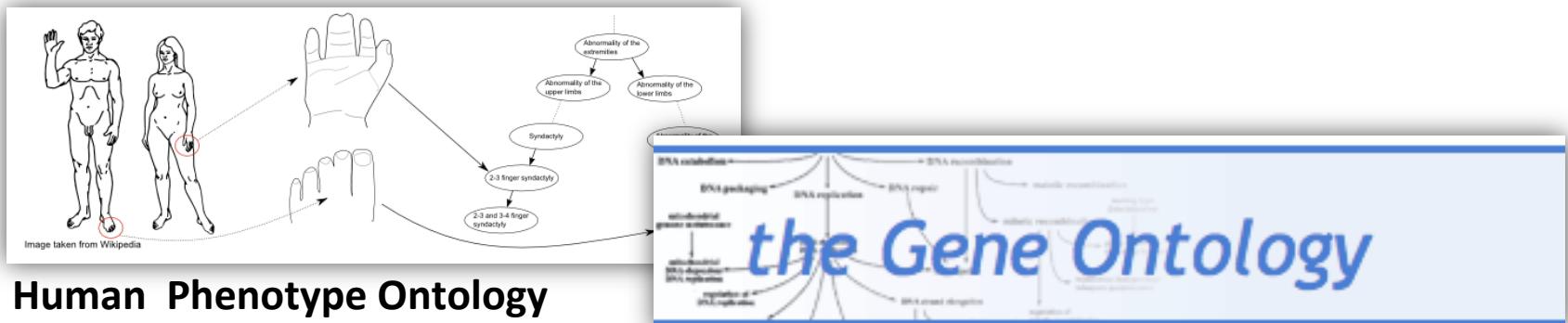
Orpha-Orpha Inferred from Gene

Knowledge Base



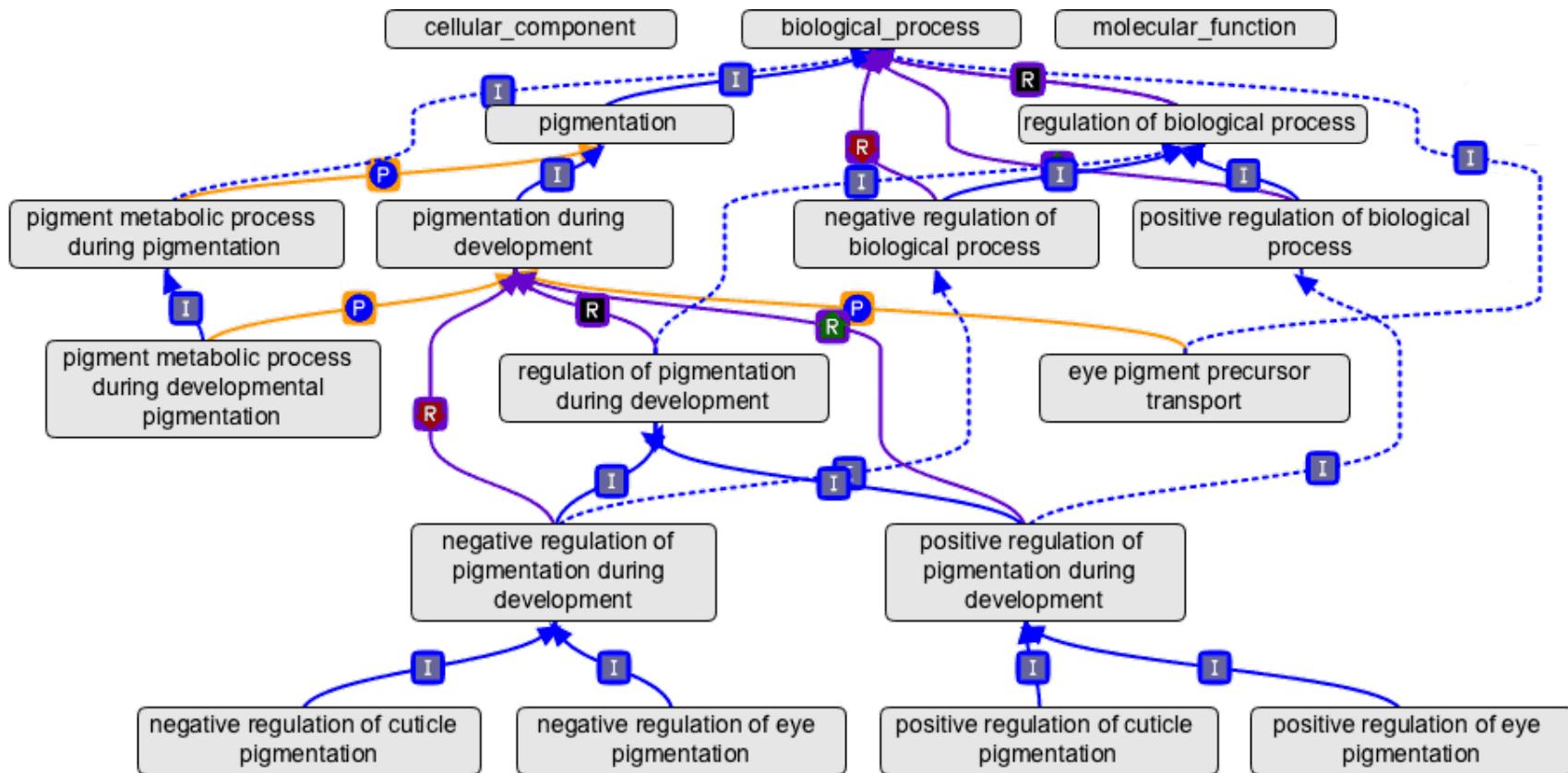
Knowledge Base : Similitud Semántica

- Similitud calculada con el uso de dos ontologías:
 - Similitud Fenotípica: Human Phenotype Ontology (HPO)
 - <http://www.human-phenotype-ontology.org/>
 - Similitud Funcional : Gene Ontology (GO)
 - <http://www.geneontology.org/>
- **Vocabularios organizados:**
 - Procesos Biológicos, Componentes Celulares, Funciones Moleculares → GO
 - Fenotipos relacionados con patologías → HPO
- **Estructura jerárquica**

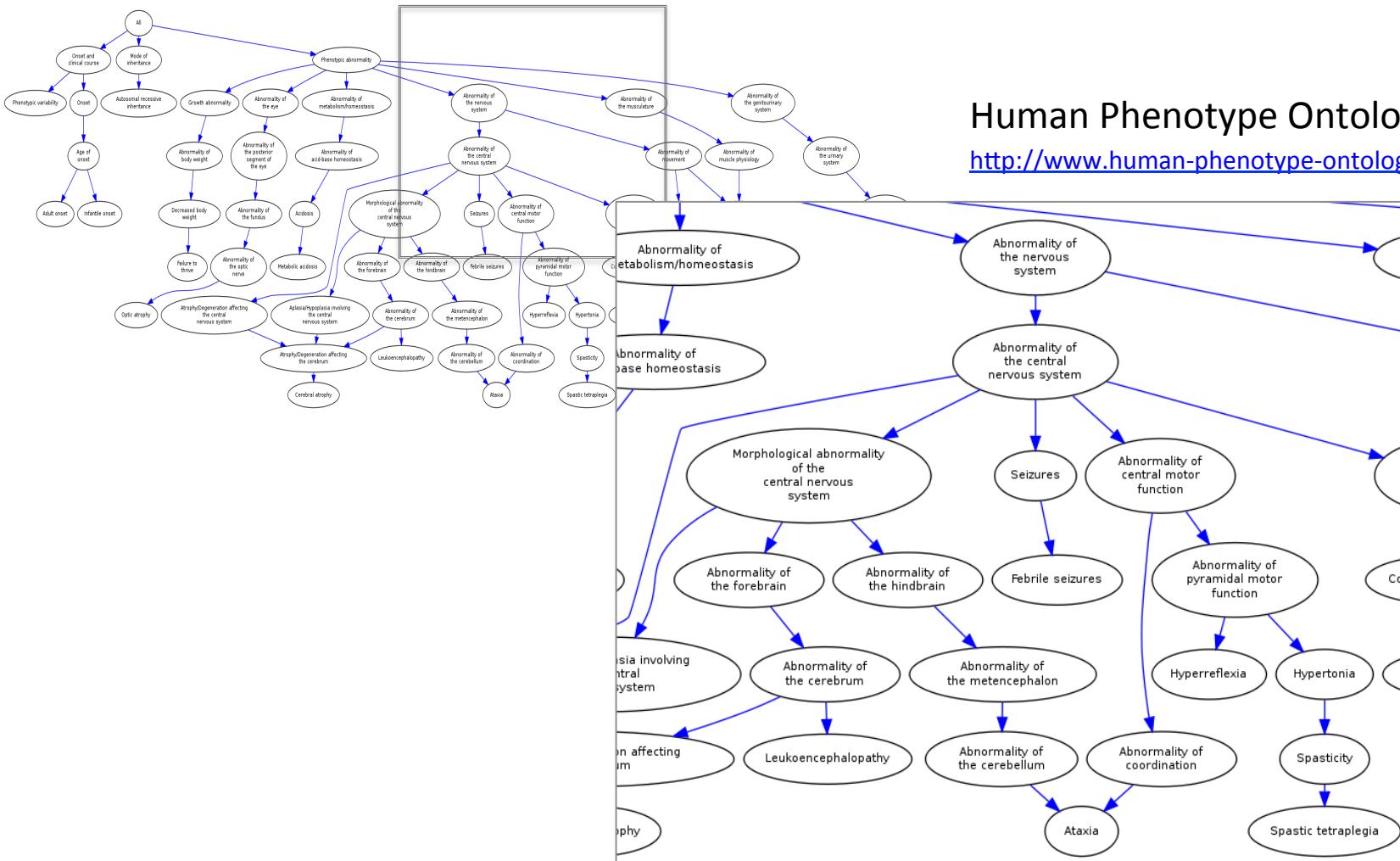


Knowledge Base : Similitud Semántica

- Gene Ontology
 - <http://www.geneontology.org/>



Knowledge Base : Similitud Semántica

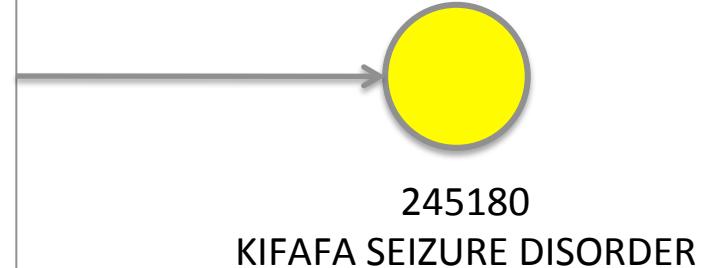


Human Phenotype Ontology
<http://www.human-phenotype-ontology.org/>

Knowledge Base : Similitud Semántica

- **Espacio fenotípico:** conjunto de fenotipos de una enfermedad a partir de la descripción de su sintomatología.

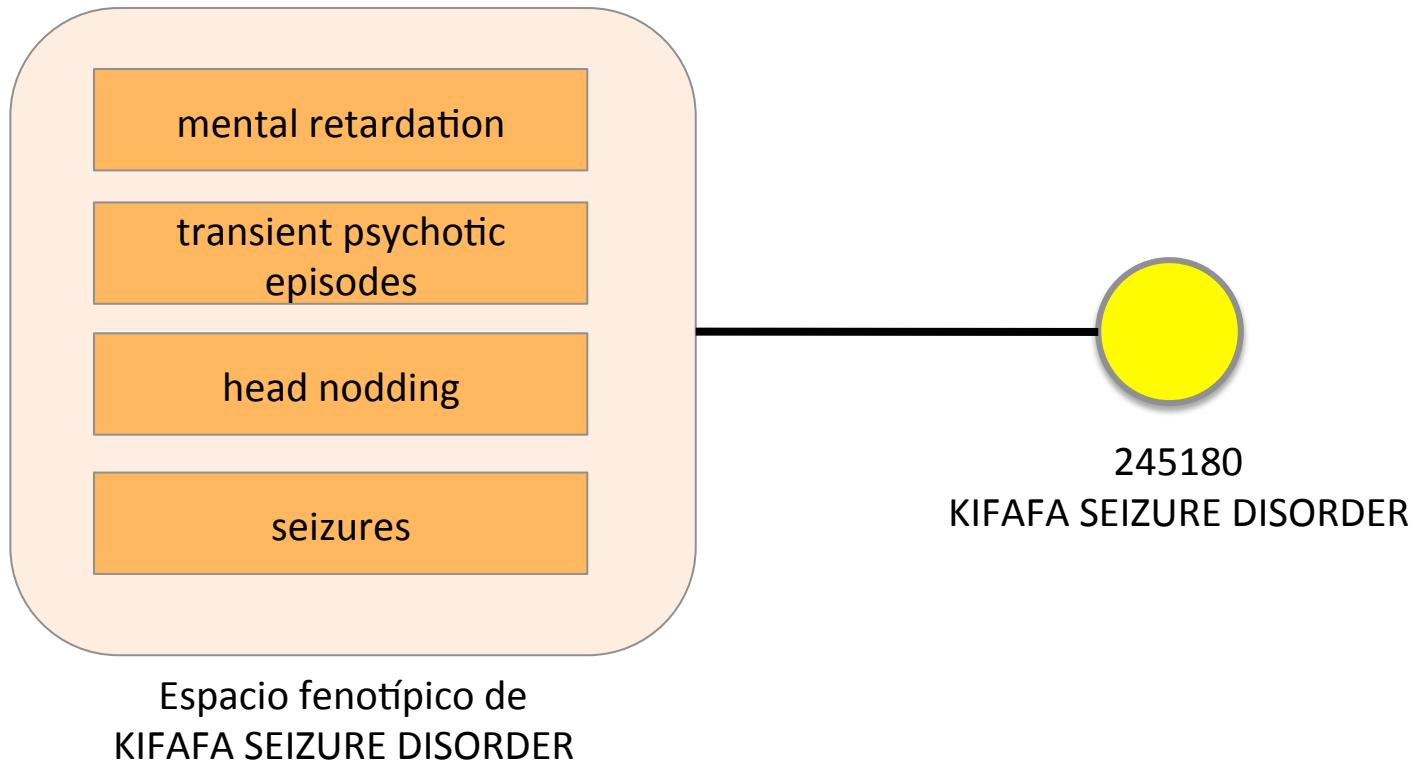
... Many showed parkinsonian features and-or other neurologic abnormalities, as well as **mental retardation** and **transient psychotic episodes**. In children, **head nodding** was a frequent precursor of later grand mal **seizures**...



<http://omim.org/entry/245180>

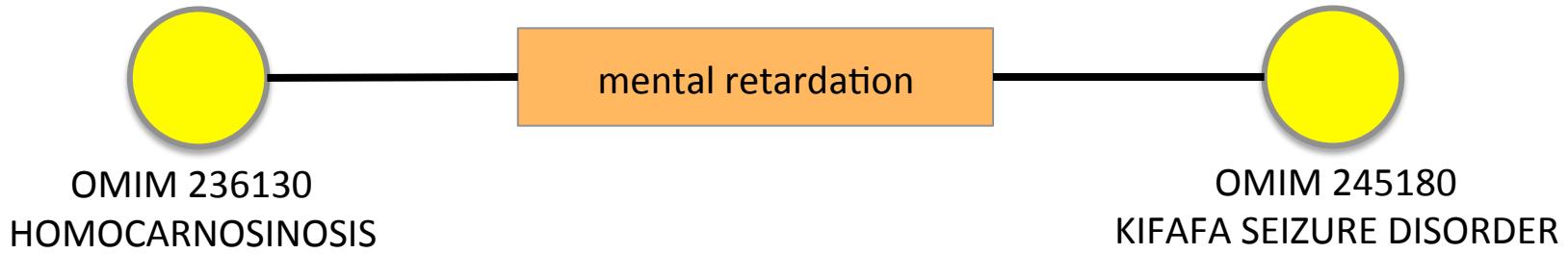
Knowledge Base : Similitud Semántica

- El vocabulario organizado permite seleccionar el conjunto de términos con los que definir la enfermedad.



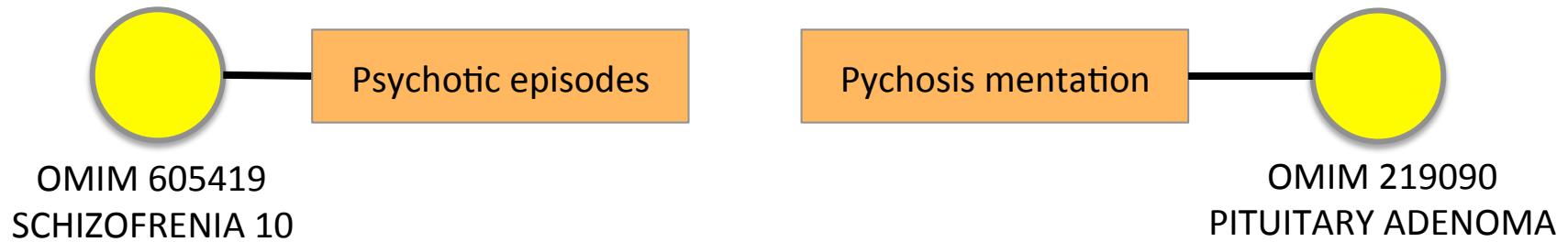
Knowledge Base : Similitud Semántica

- Relaciones entre enfermedades a partir de los fenotipos.
- Similar a la relaciones inferidas.



Knowledge Base : Similitud Semántica

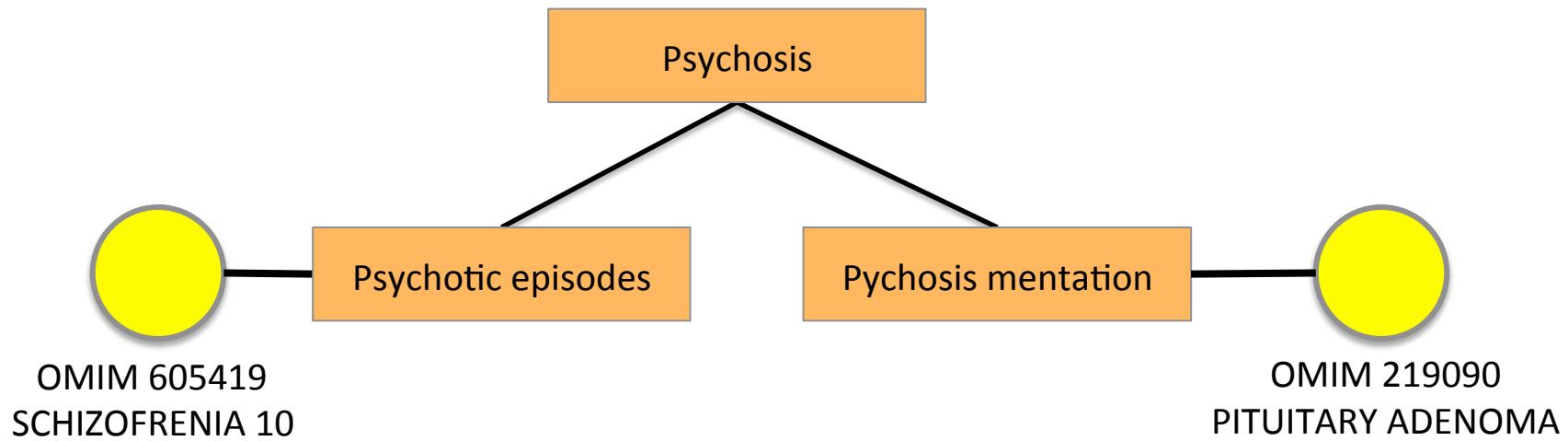
- La estructura jerárquica de la ontología permite ampliar las relaciones que pueden establecerse a partir de inferencias.



¿Deberían estar relacionadas?

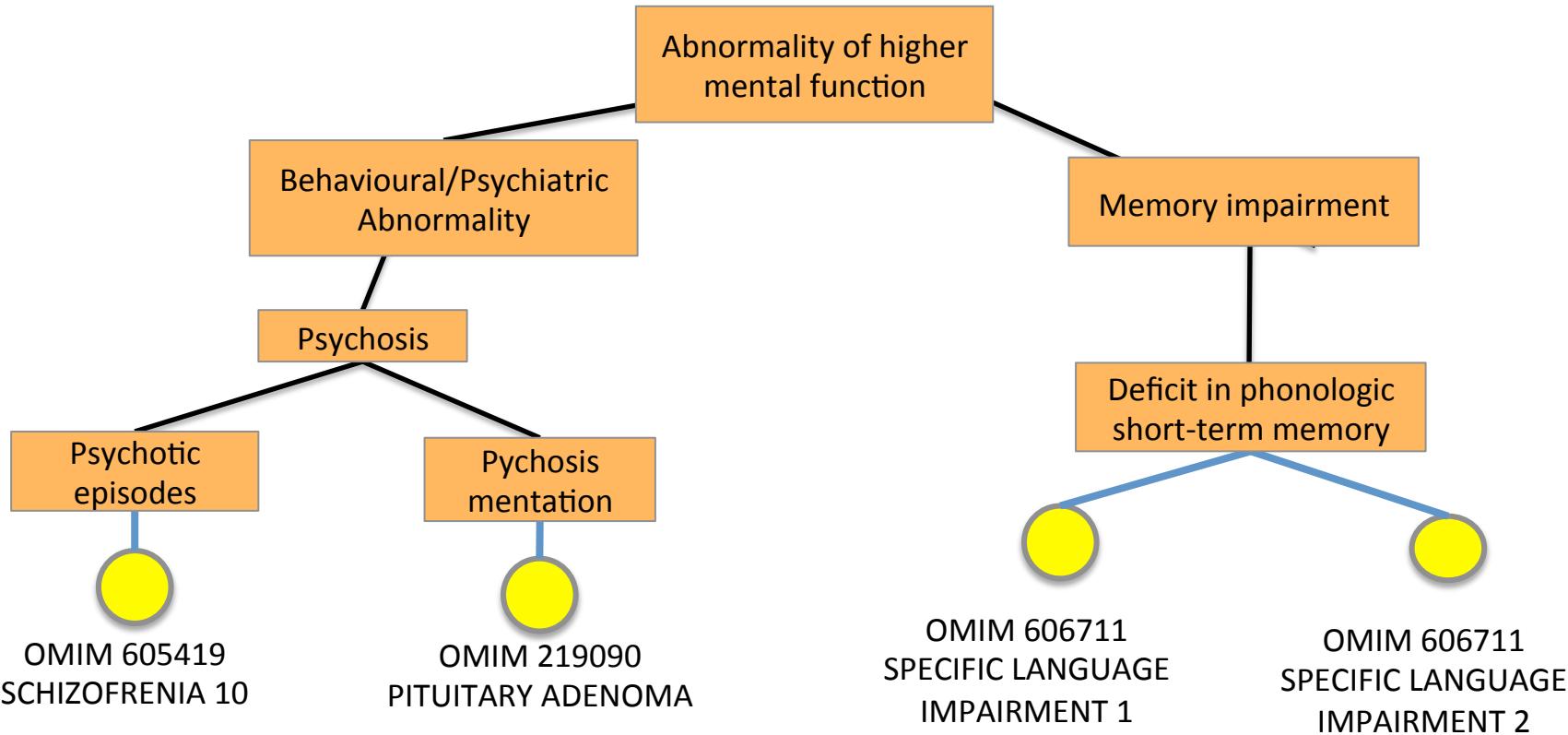
Knowledge Base : Similitud Semántica

- El fenotipo “pychosis” permite establecer una relación entre las dos enfermedades.



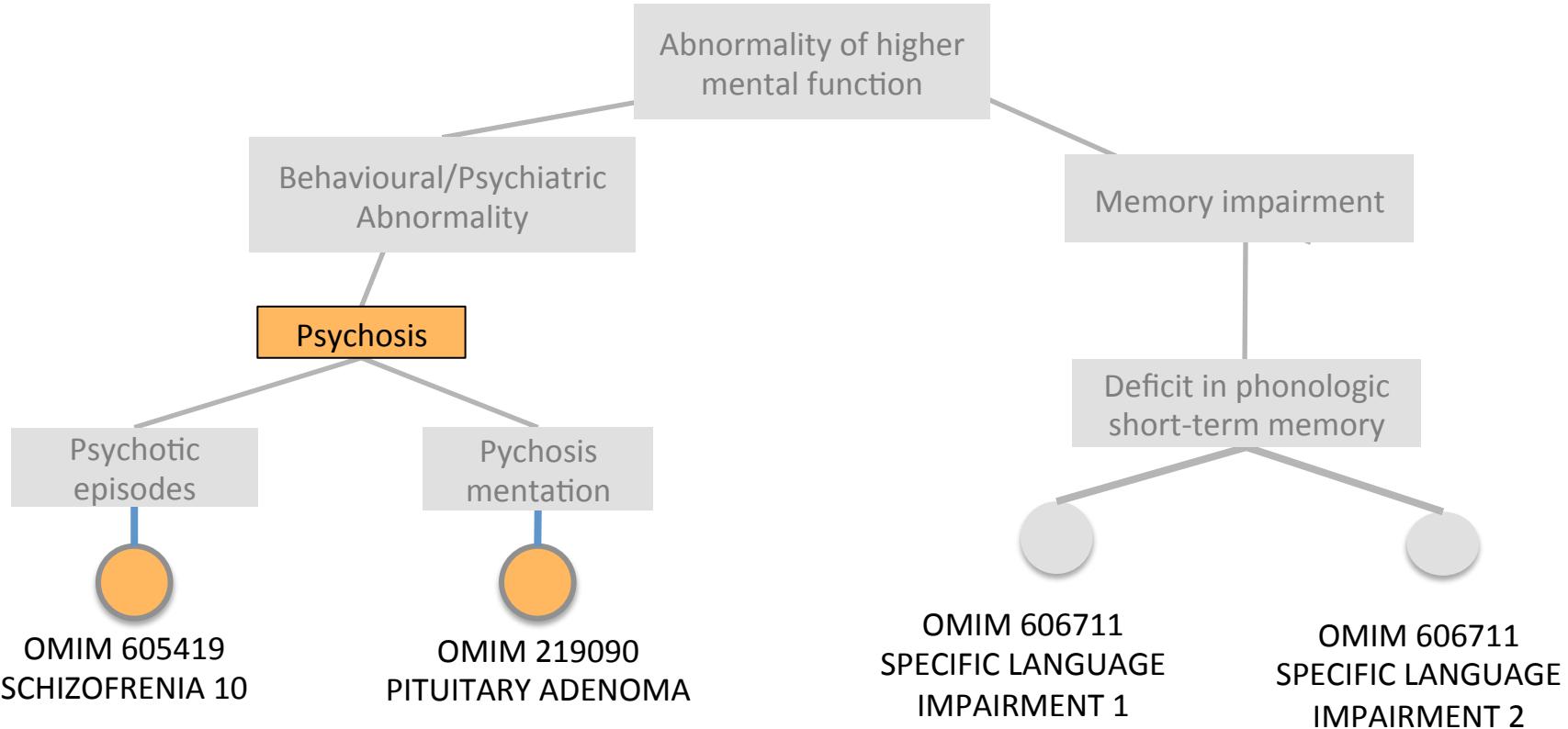
Knowledge Base : Similitud Semántica

- La lejanía del término común entre los síntomas de dos enfermedades van a determinar el valor de la similitud semántica.



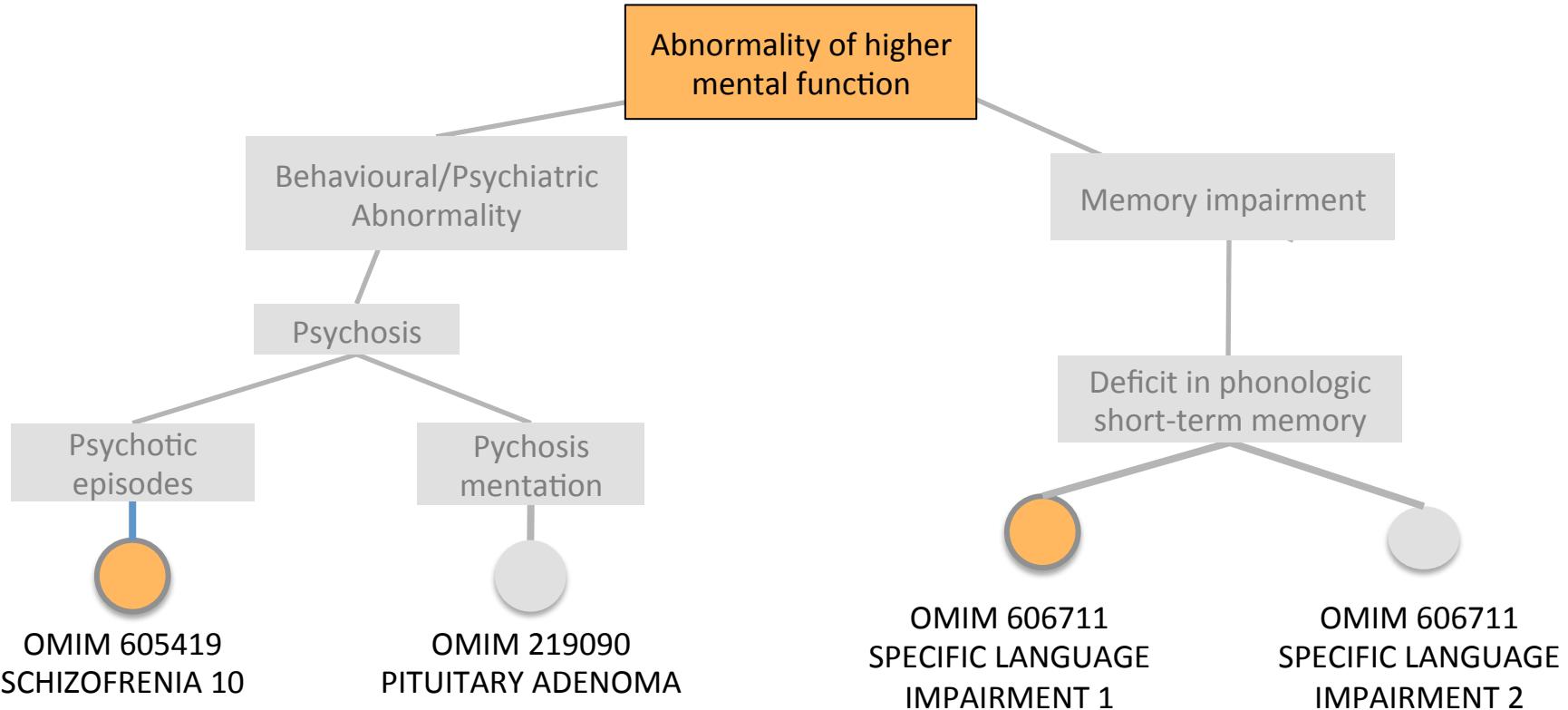
Knowledge Base : Similitud Semántica

- La similitud entre **Schizofrenia** y **Pituitary adenoma** será mayor que la similitud entre **Schizofrenia** y **Specific Language Impairment 1**.



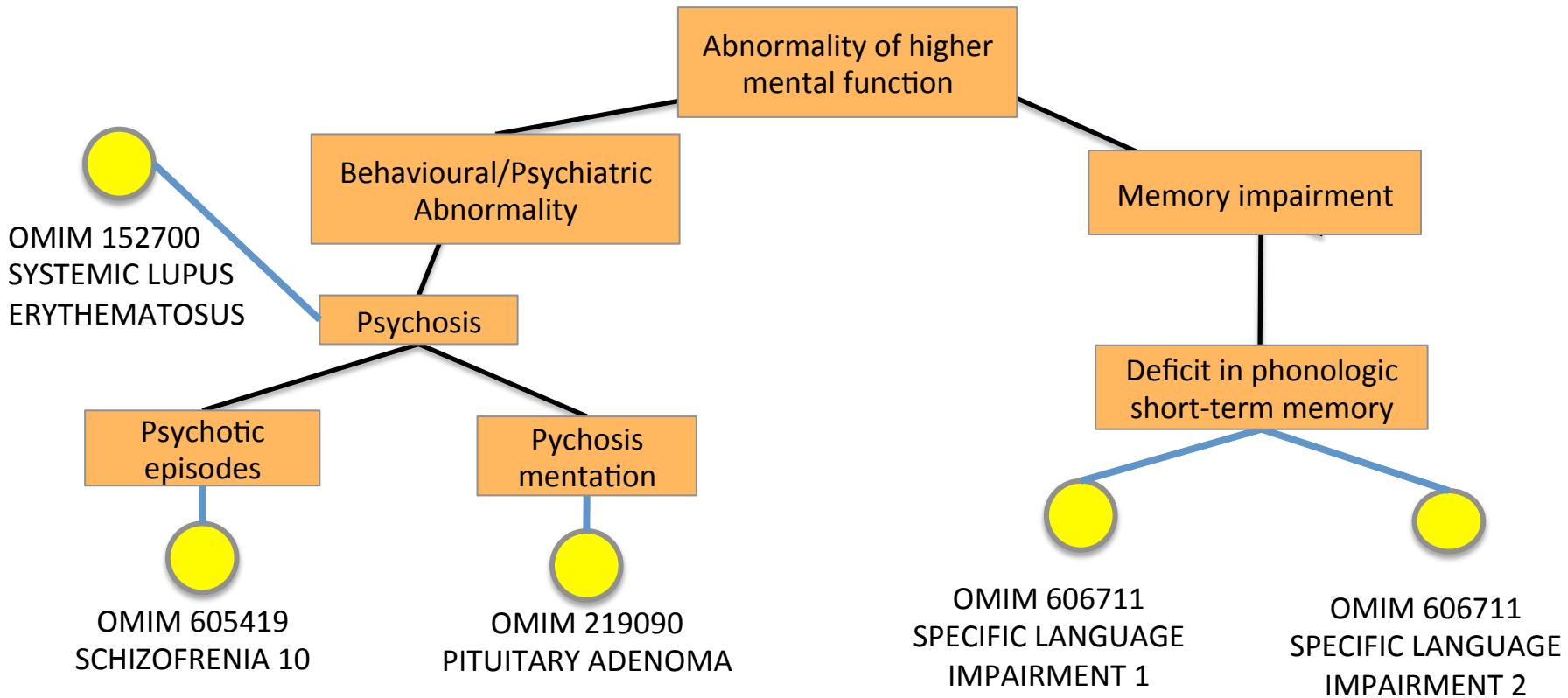
Knowledge Base : Similitud Semántica

- La similitud entre **Schizofrenia** y **Pituitary adenoma** será mayor que la similitud entre **Schizofrenia** y **Specific Language Impairment 1**.



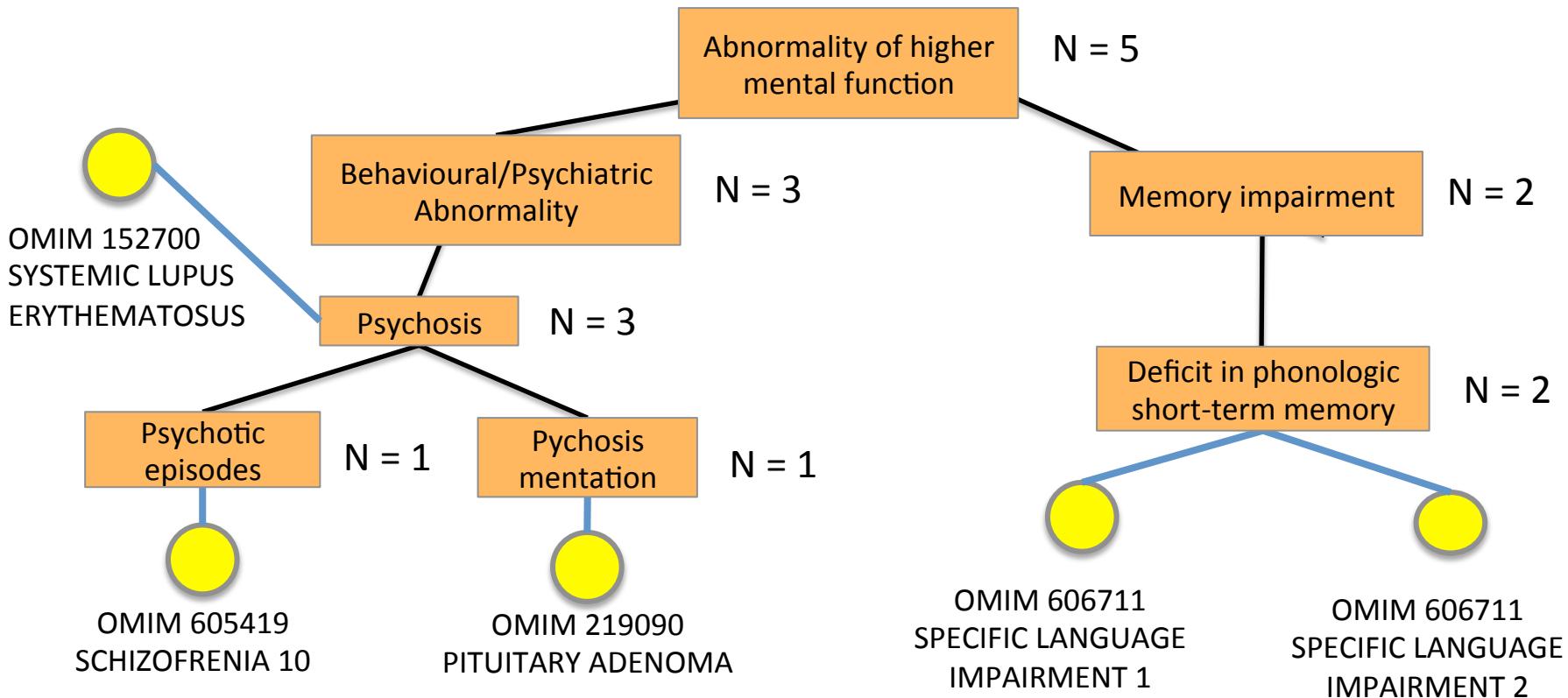
Knowledge Base : Similitud Semántica

- A cada fenotipo del árbol se le asigna un valor numérico (IC) que será mayor a medida que nos alejamos de la raíz.
- Relaciones fenotipo – enfermedad se propagan hacia la raíz



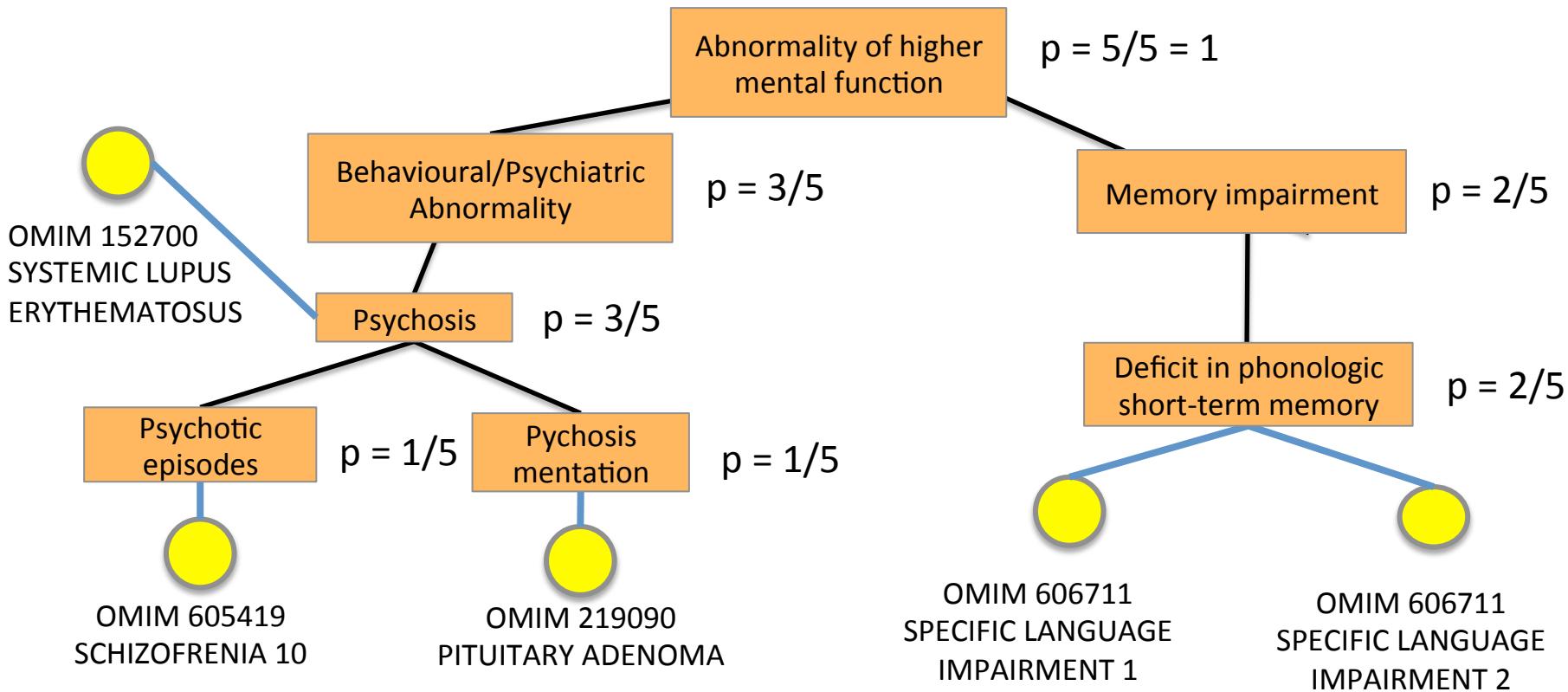
Knowledge Base : Similitud Semántica

- N = número de enfermedades asociadas a un fenotipo



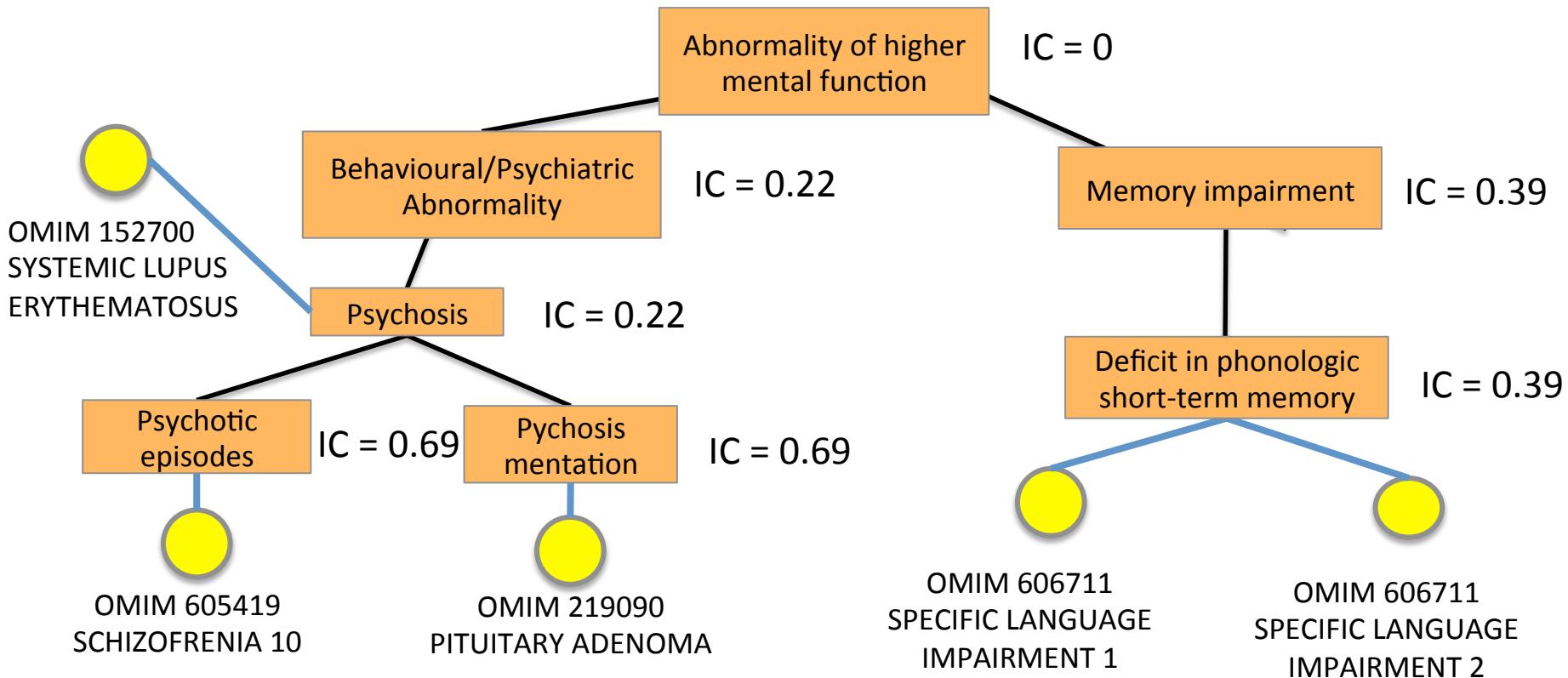
Knowledge Base : Similitud Semántica

- Probabilidad de cada término $p(t)$



Knowledge Base: Similitud Semántica

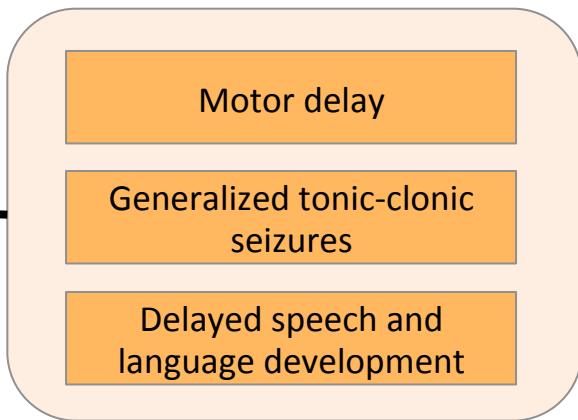
- $IC = - \log(p(t))$



Knowledge Base : Similitud Semántica

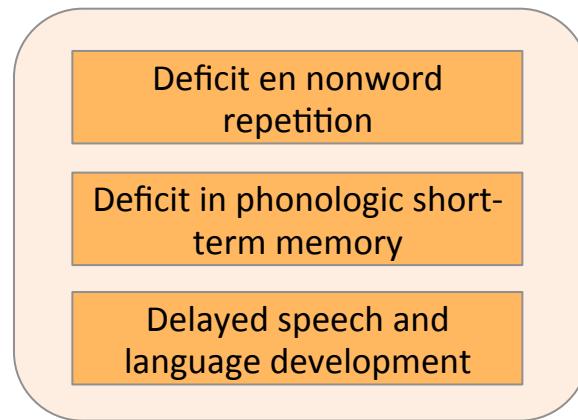
- Una enfermedad no se define por un solo fenotípico sino por un conjunto de fenotipos.

OMIM 300423
MENTAL RETARDATION



Espacio fenotípico de
MENTAL RETARDATION

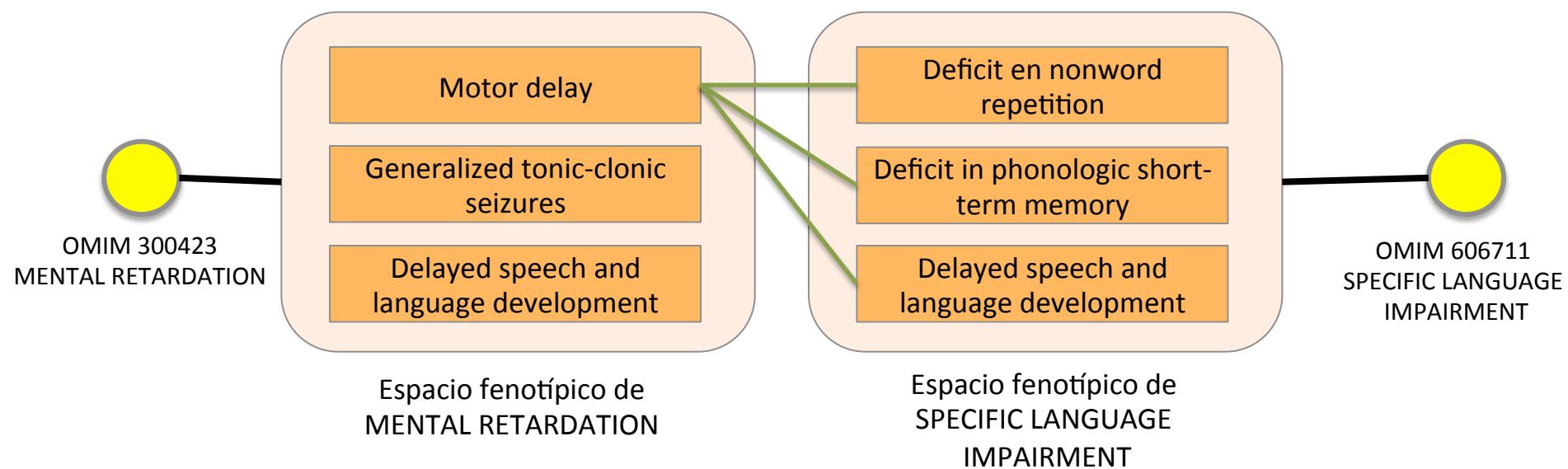
OMIM 606711
SPECIFIC LANGUAGE
IMPAIRMENT



Espacio fenotípico de
SPECIFIC LANGUAGE
IMPAIRMENT

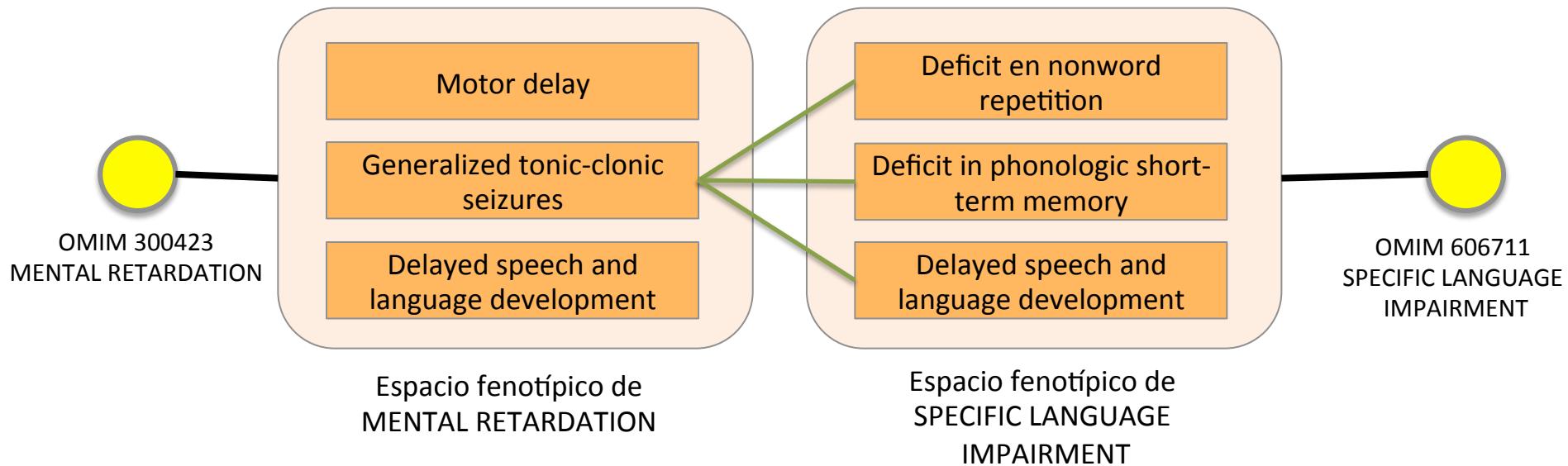
Knowledge Base : Similitud Semántica

- Para comparar dos grupos de fenotipos se comparan uno a uno los fenotipos de las dos enfermedades.



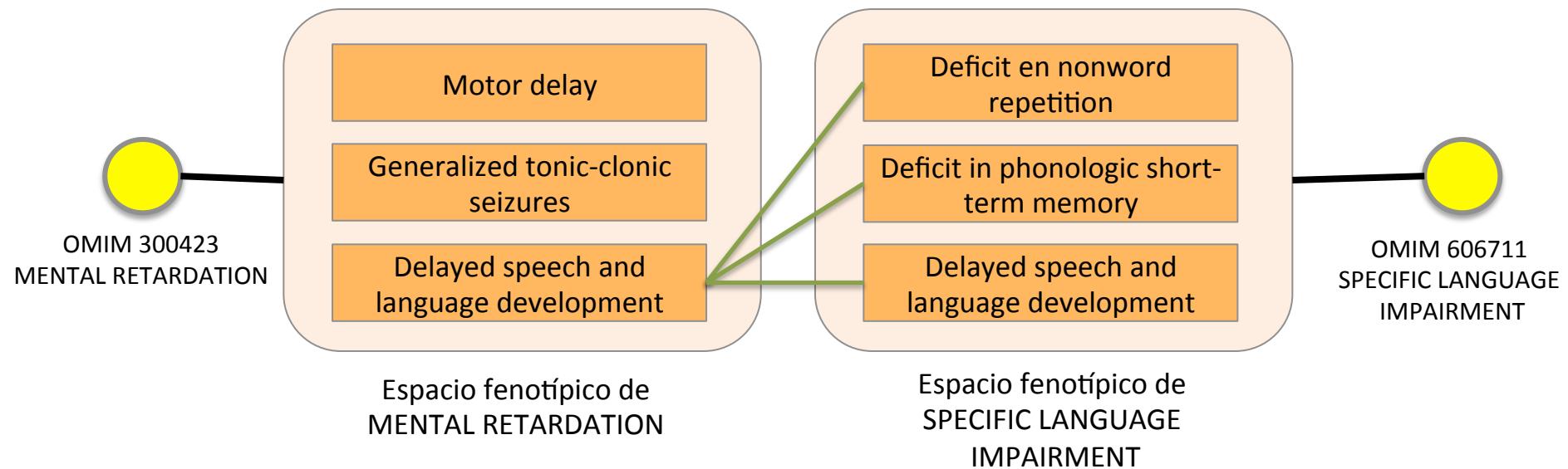
Knowledge Base : Similitud Semántica

- Para comparar dos grupos de fenotipos se comparan uno a uno los fenotipos de las dos enfermedades.



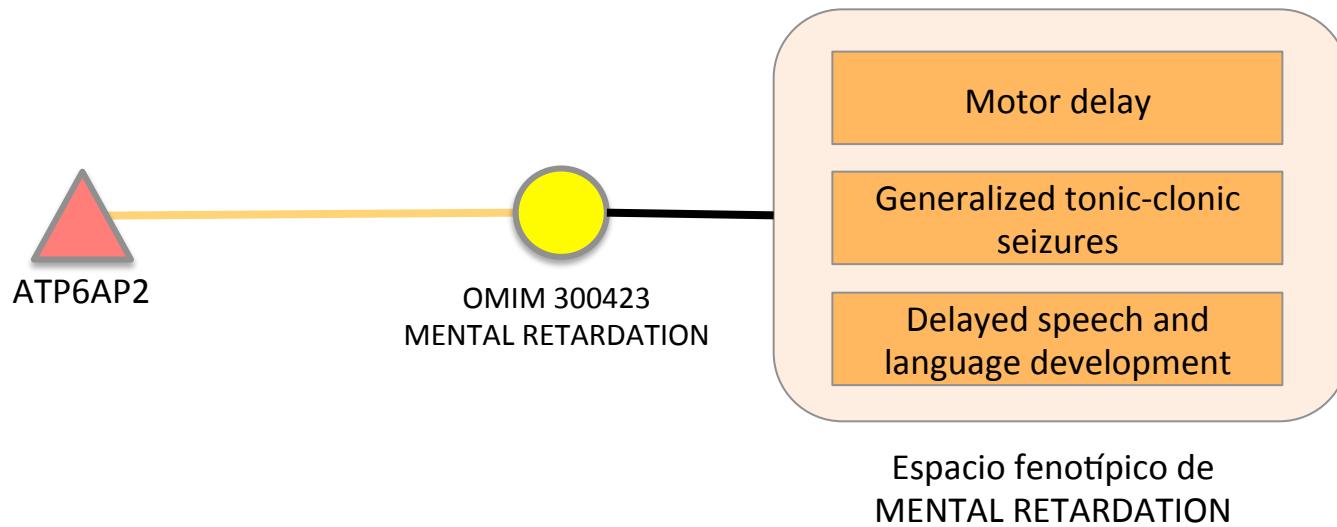
Knowledge Base : Similitud Semántica

- Para comparar dos grupos de fenotipos se comparan uno a uno los fenotipos de las dos enfermedades.
- El valor de similitud fenotípica entre las dos enfermedades es el **promedio de todas estas comparaciones**



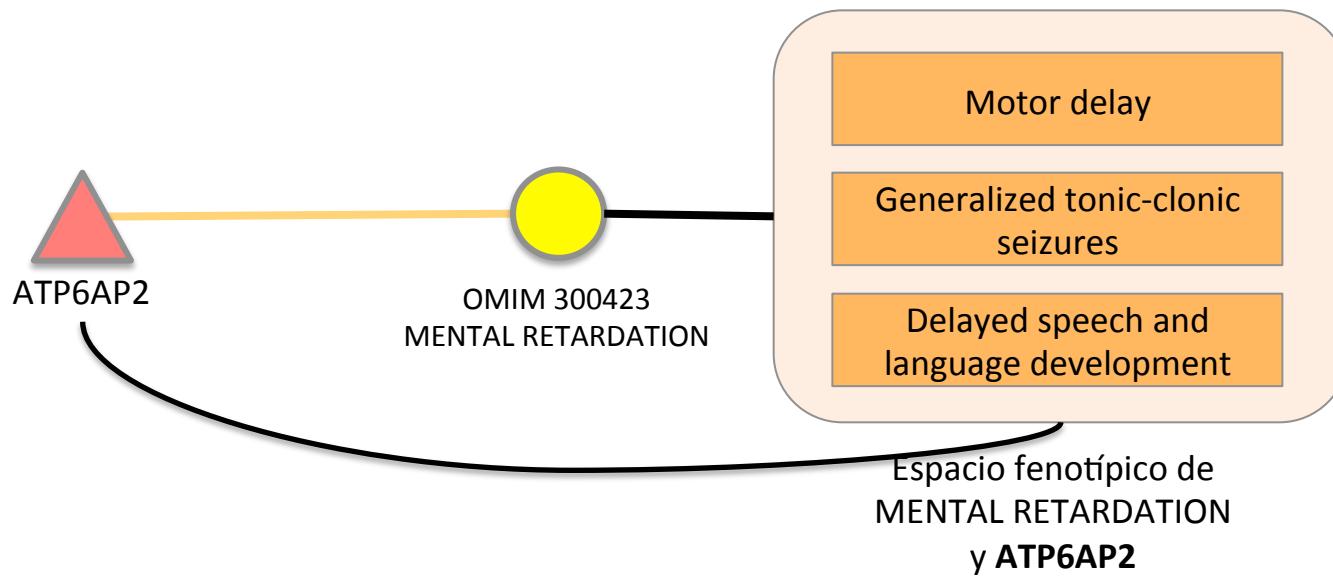
Knowledge Base : Similitud Semántica

- A partir Orphanet también es posible definir un conjunto de fenotipos a enfermedades raras.
- Las relaciones entre un conjunto de fenotipos y los genes se establecen a partir de las relaciones conocidas: gen-omim.



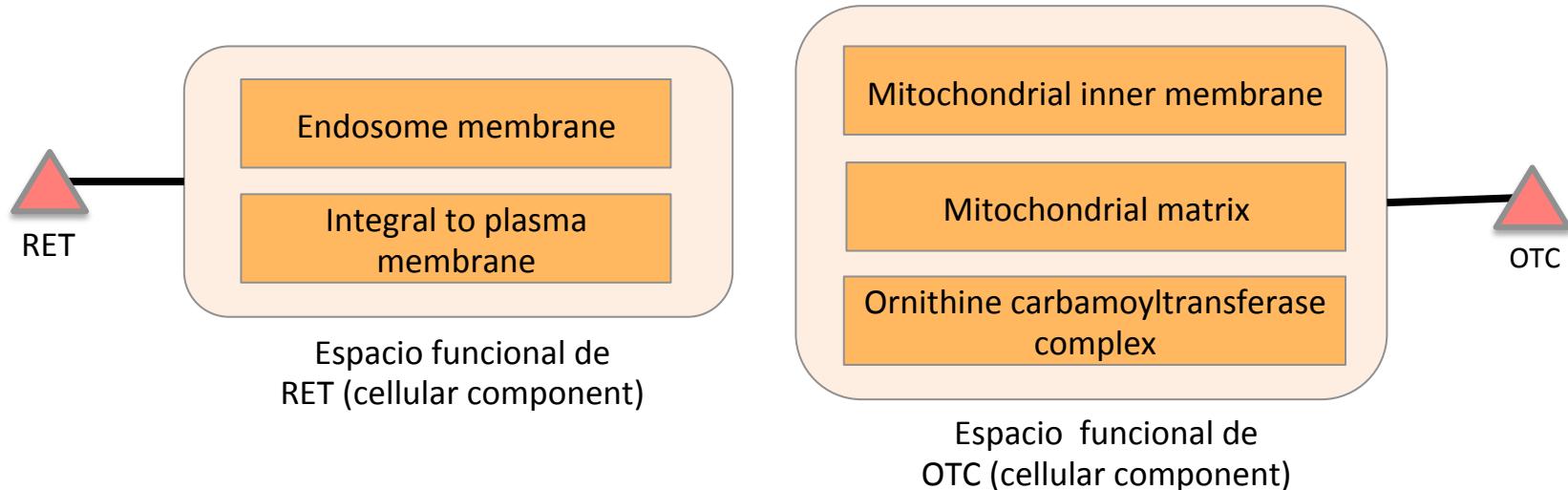
Knowledge Base : Similitud Semántica

- A partir de la información de **Orphanet** también es posible definir un conjunto de fenotipos a **enfermedades raras**.
- Las relaciones entre un conjunto de fenotipos y un **gen** se establecen a partir de las **relaciones conocidas**: gen-omim.



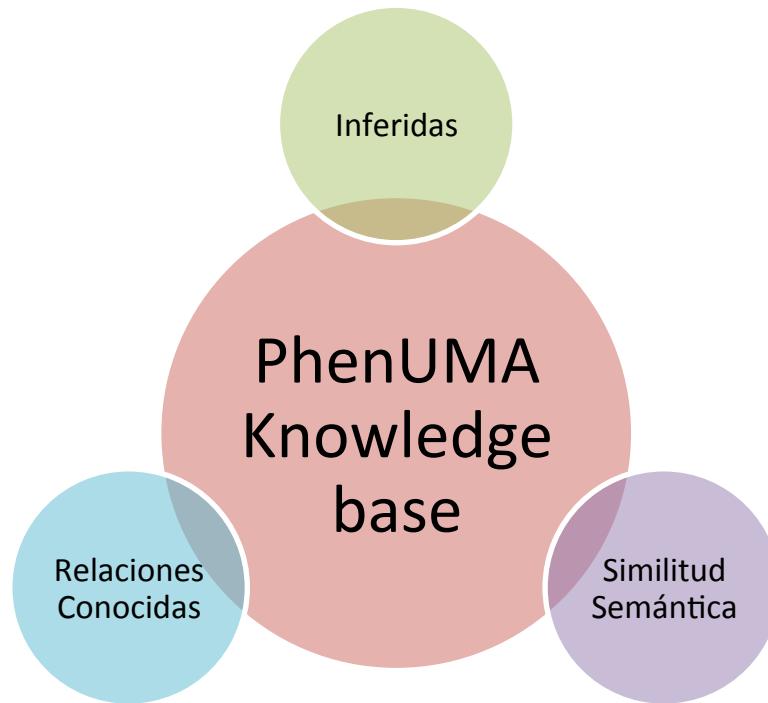
Knowledge Base : Similitud Semántica

- El cálculo de la similitud semántica funcional entre **genes** es similar en el caso de la Gene Ontology (GO).
- El lugar de fenotipos patológicos los genes se relacionan a partir de **procesos biológicos, componentes celulares y funciones moleculares**.



Knowledge Base : Similitud Semántica

- A partir de los espacios fenotípicos y funcionales se han creado estos tipos de redes de similitud semántica:



OMIM-OMIM SimSem from HPO
ORPHA-ORPHA SimSem from HPO
Gene-Gene SimSem from HPO

Gene-Gene SimSem from GO (Biological Process)
Gene-Gene SimSem from GO (Cellular Component)
Gene-Gene SimSem from GO (Molecular Function)

Tipos de Redes

Entrada	Total Relaciones	
Genes	Gene-Gene Known Protein-Protein Interaction from STRING	Conocidas
	Gene-Gene Known Metabolic Interaction (Veeramani et al.)	
	Gene-OMIM Known from OMIM	
	Gene-ORPHA Known from Orphanet	
	Gene-Gene Inferred from OMIM	Inferidas
	Gene-Gene Inferred from Orphanet	
	Gene-Gene SimSem from HPO	
	Gene-Gene SimSem from GO (Biological Process)	Similitud Semántica
	Gene-Gene SimSem from GO (Cellular Component)	
	Gene-Gene SimSem from GO (Molecular Function)	
	ORPHA-ORPHA SimSem from HPO	
	OMIM-OMIM SimSem from HPO	

Tipos de Redes

Entrada	Tipos de Redes	
	Gene-OMIM Known from OMIM	Conocidas
	OMIM-OMIM Inferred from Genes	Inferidas
Enfermedades OMIM	Gene-Gene SimSem from HPO	
	Gene-Gene SimSem from GO (Biological Process)	
	Gene-Gene SimSem from GO (Cellular Component)	
	Gene-Gene SimSem from GO (Molecular Function)	
	OMIM-OMIM SimSem from HPO	

Tipos de Redes

Entrada	Tipos de Redes	
	Gene-ORPHAKnown from OMIM	Conocidas
	ORPHA-ORPHAIInferred from Genes	Inferidas
Enfermedades Orphanet	Gene-Gene SimSem from HPO	
	Gene-Gene SimSem from GO (Biological Process)	
	Gene-Gene SimSem from GO (Cellular Component)	
	Gene-Gene SimSem from GO (Molecular Function)	
	ORPHA-ORPHA SimSem from HPO	

Tipos de Redes

- En consultas de fenotipos las redes de salida consisten en el conjunto de genes, enfermedades OMIM o enfermedades raras (Orphanet) que más se parece al conjunto de fenotipos de entrada.



Nivel de Confianza

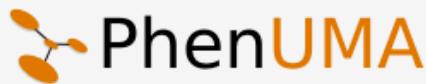
- Cortes y Niveles de Confianza

Red	Total Relaciones	Relaciones TOP 2% (Low)	Relaciones TOP 1%(Medium)	Relaciones TOP 0.5% (High)
ORPHA-ORPHA SimSem from HPO	3804649	75924	37702	18875
OMIM-OMIM SimSem from HPO	7500235	149689	74935	37483
Gene-Gene SimSem from HPO	1245519	24902	12437	6216

Red	Total Relaciones	Relaciones TOP 0.5% (Low)	Relaciones TOP 0.2% (Medium)	Relaciones TOP 0.1%(High)
Gene-Gene SimSem from GO (Biological Process)	99073951	486982	198621	95023
Gene-Gene SimSem from GO (Cellular Component)	113426652	565739	226854	112115
Gene-Gene SimSem from GO (Molecular Function)	80118330	397683	169626	80391

Construcción de Redes

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PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases

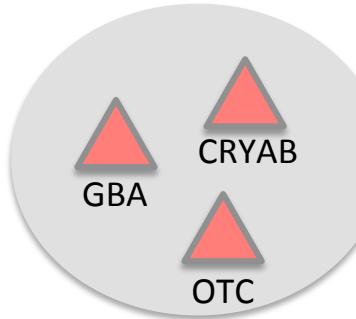
Type of Input: ID: Type of Output Network: Confidence:

Input List:

Construcción de la Red

Construcción de Redes

1. Genes de Interés



¿Con que otros genes se relacionan mis genes de interés fenotípicamente?

2. Tipo de Red de Salida:

Ej: Similitud Fenotípica con otros genes
(Gene-Gene SimSim from HPO)

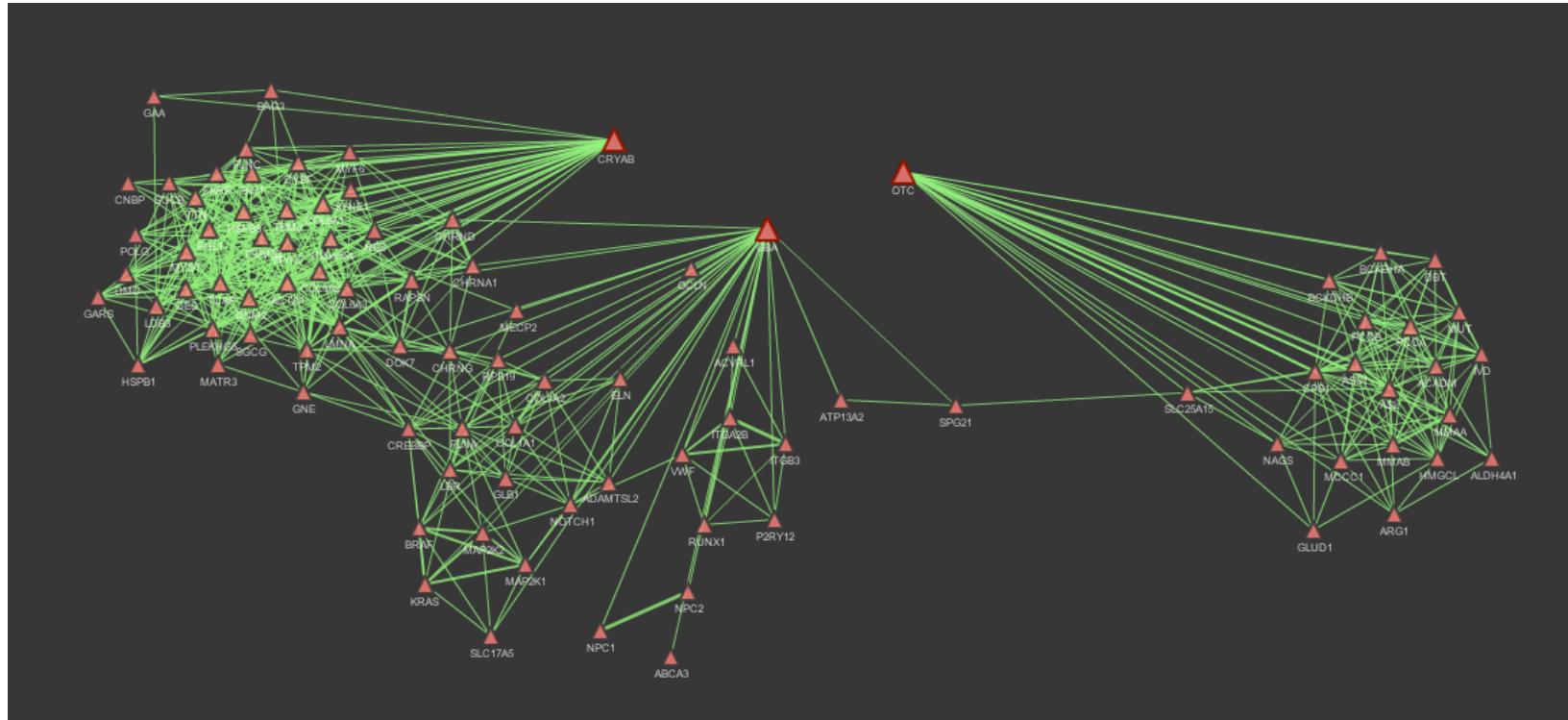
3. Nivel de Confianza

Ej: Low

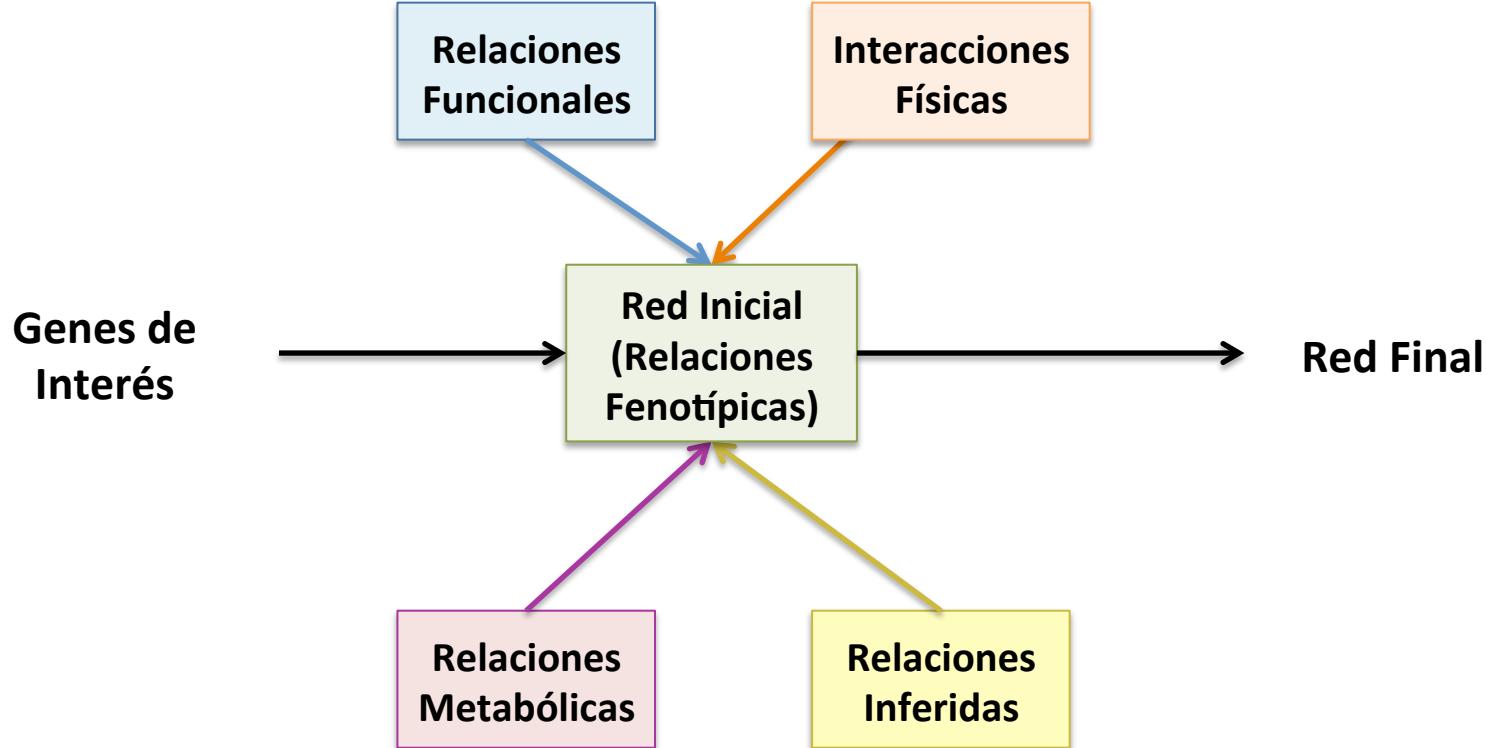
Red Inicial
(Relaciones Fenotípicas)

Construcción de Redes

- **Red Inicial:** Tipo de relación seleccionada (relaciones fenotípicas)



Knowledge Base : Construcción de Redes



Construcción de Redes

- **Red Final:** Enriquecida con el resto de relaciones entre los genes de la red resultante

