

Sesión Práctica I: Introducción a



PhenUMA

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




UNIVERSIDAD
DE MÁLAGA



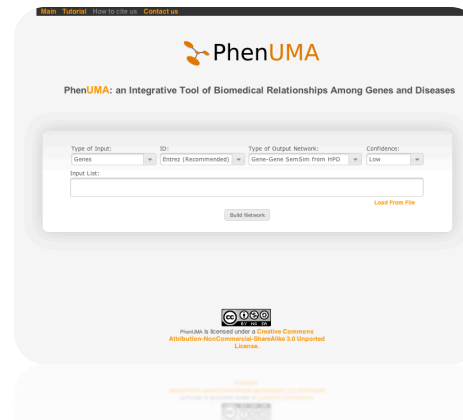
Sesión Práctica: Esquema



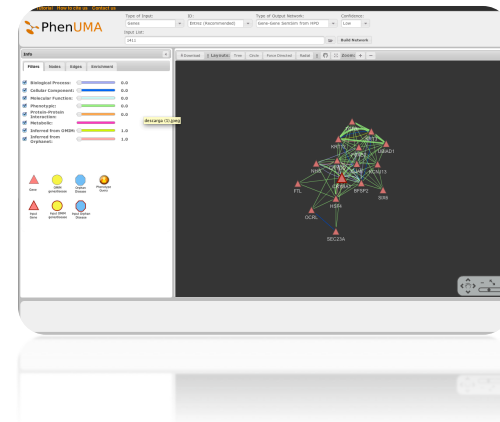
- **Sesión Práctica I:**
 - Entradas de Datos
 1. Opciones de Entrada
 2. Datos de Entrada
 3. Opciones de la Red de Salida
 - Knowledge Base
 - Tipos de Redes
 - Niveles de Confianza
 - Construcción de Redes
 - Funcionalidades de la Aplicación
 - **Sesión Práctica II:**
 - Gestión de los Datos de Salida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

Interfaz de PhenUMA

- Página Principal



- Página de Visualización



Entrada a PhenUMA

1. Opciones de Entrada

3. Opciones de la Red de Salida

The screenshot shows the PhenUMA web interface. At the top, there is a navigation bar with links for 'Main', 'Tutorial', 'How to cite us', and 'Contact us'. Below this is the PhenUMA logo and the title 'PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases'. The main form contains several input fields and a button. A red box highlights the 'Type of Input' and 'ID' dropdowns, with a red arrow pointing to the 'Type of Input' dropdown. A green box highlights the 'Type of Output Network' and 'Confidence' dropdowns, with a green arrow pointing to the 'Type of Output Network' dropdown. A blue box highlights the 'Input List' text area, with a blue arrow pointing to it. A 'Load From File' link is located to the right of the 'Input List' field. A 'Build Network' button is at the bottom of the form.

Main Tutorial How to cite us Contact us

PhenUMA

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/>

Orphanum

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

Indetificadores de Genes: NCBI

The image shows a screenshot of the NCBI (National Center for Biotechnology Information) website. The top navigation bar includes the NCBI logo, 'Resources' (checked), 'How To' (checked), and a 'Sign in to NCBI' link. A search bar is located in the top right. The left sidebar contains a 'Resource List (A-Z)' with categories like 'All Resources', 'Chemicals & Bioassays', 'Data & Software', 'DNA & RNA', 'Domains & Structures', 'Genes & Expression', 'Genetics & Medicine', 'Genomes & Maps', 'Homology', 'Literature', 'Proteins', 'Sequence Analysis', 'Taxonomy', 'Training & Tutorials', and 'Variation'. A dropdown menu is open over the 'Gene' category, listing various databases and tools such as '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', and 'UniSTS'. The main content area features a 'Welcome to NCBI' message, a 'Popular Resources' section with links to PubMed, Bookshelf, PubMed Central, PubMed Health, BLAST, Nucleotide, Genome, SNP, Gene, Protein, and PubChem, and an 'NCBI Announcements' section with recent news items. A 'Twitter feed' widget is also visible.

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

Identificadores de Genes: NCBI

Entrez/Gene ID

NCBI Resources How To Sign in to NCBI

Gene Search Limits Advanced Help

Display Settings: Full Report Send to:

CRYBA1 crystallin, beta A1 [*Homo sapiens* (human)]

Gene ID: 1411, updated on 9-Jun-2013

Summary

Official Symbol CRYBA1 provided by HGNC

Official Full Name crystallin, beta A1 provided by HGNC

Primary source HGNC:2394

See related [Ensembl:ENSG00000108255](#); [HPRD:00429](#); [MIM:123610](#); [Vega:OTTHUMG00000132729](#)

Gene type protein coding

RefSeq status REVIEWED

Organism [Homo sapiens](#)

Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorhini; Catarrhini; Hominidae; Homo

Also known as CRYB1; CTRCT10

Summary Crystallins are separated into two classes: taxon-specific, or enzyme, and ubiquitous. The latter class constitutes the major proteins of vertebrate eyes, and maintains the transparency and refractive index of the lens. Since lens central fiber cells lose their nuclei during development, these crystallins are made and then retained throughout life, making them extremely stable proteins. Mammalian lens crystallins are divided into alpha, beta, and gamma families; beta and gamma crystallins are also considered as a superfamily. Alpha and beta families are further divided into acidic and basic groups. Seven protein regions exist in crystallins: four homologous motifs, a connecting peptide, and N- and C-terminal extensions. Beta-crystallins, the most heterogeneous, differ by the presence of the C-terminal extension (present in the basic group, none in the acidic group). Beta-crystallins form aggregates of different sizes and are able to self-associate to form dimers or to form heterodimers with other beta-crystallins. This gene, a beta acidic group member, encodes two proteins (crystallin, beta A3 and crystallin, beta A1) from a single mRNA, the latter protein is 17 aa shorter than crystallin, beta A3 and is generated by use of an alternate translation initiation site. Deletion of exons 3 and 4 causes the autosomal dominant disease 'zonular cataract with sutural opacities'. [provided by RefSeq, Jul 2008]

Table of contents

- Summary
- Genomic context
- Genomic regions, transcripts, and products
- Bibliography
- Phenotypes
- Interactions
- General gene information
 - Markers, Homology, Gene Ontology
- General protein information
- Reference sequences
- Related sequences
- Additional links

Related information

- Order cDNA clone
- BioAssay
- BioProjects
- CCDS
- ClinVar
- Conserved Domains
- dbVar
- EST
- Full text in PMC

Official Symbol

Más Identificadores

Indetificadores de Genes: Ensembl

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

The screenshot shows the Ensembl genome browser interface. At the top, there is a navigation bar with the Ensembl logo and links for BLAST/BLAT, BioMart, Tools, Downloads, Help & Documentation, Blog, and Mirrors. A search bar on the right contains the text "Search all species...". Below the navigation bar, a search box is pre-filled with "Human" and "Beta-crystallin A3", with a "Go" button. Below the search box, there are suggestions: "e.g. BRCA2 or rat X:100000..200000 or coronary heart disease".

The main content area is divided into several sections:

- Browse a Genome:** A section describing the Ensembl project's goal of producing genome databases for vertebrates and other eukaryotic species.
- Popular genomes:** A list of popular genomes with icons and labels: Human (GRCm37), Mouse (GRCm38), and Zebrafish (Zv9). A link "Log in to customize this list" is provided.
- All genomes:** A dropdown menu to "Select a species" and a link to "View full list of all Ensembl species".
- Other species:** A note that other species are available in "Ensembl Pre!" and "EnsemblGenomes".
- ENCODE data in Ensembl:** A section with the ENCODE logo and a DNA double helix icon.
- Variant Effect Predictor (VeP):** A section with the VeP logo.
- New to Ensembl?:** A list of resources for new users:
 - [Learn how to use Ensembl](#) with video tutorials and walk-throughs.
 - [Add custom tracks](#) using the control panel.
 - [Upload and analyse your data](#) and save it to your Ensembl account.
 - [Search for a DNA or protein sequence](#) using BLAST or BLAT.
 - [Fetch only the data you want](#) from the public database using the Perl API.
 - [Download our databases via FTP](#) in FASTA, MySQL, and other formats.
 - [Mine Ensembl with BioMart](#) and export sequences or tables in text, HTML, or Excel format.
- Still got questions?** A link to "FAQs" or "glossary".
- What's New in Release 71 (April 2013):** A list of recent updates:
 - [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** and **More release news on our blog** links.
- Latest blog posts:** A list of recent 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** link.
- Did you know...?:** A tip about adjusting the display width, with an icon of a monitor and a "Display options" menu.

Indetificadores 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

Ensembl BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors Login/Register

Human (GRCh37) Search Human...

Search Ensembl
New Search
Configure this page
Add your data
Export data
Bookmark this page
Share this page

Results Summary

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

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

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

Ensembl release 71 - April 2013 © [WTSI](#) / [EBI](#) [About Ensembl](#) | [Privacy Policy](#) | [Contact Us](#)

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

Indetificadores de Genes: Ensembl

The screenshot shows the Ensembl genome browser interface. At the top, the Ensembl logo is on the left, and navigation links (BLAST/BLAT, BioMart, Tools, Downloads, Help & Documentation, Blog, Mirrors) and a search bar are on the right. Below the navigation bar, the 'Human (GRCh37)' dropdown is selected. On the left side, there are several utility buttons: 'Search Ensembl' (with a 'New Search' link), 'Configure this page', 'Add your data', 'Export data', 'Bookmark this page', and 'Share this page'. The main content area is titled 'Result in Detail' and shows '1 Gene matches your query ('Beta-crystallin A3') in Human'. The gene 'CRYBA1' is listed with the following details:

Description	crystallin, beta A1 [Source:HGNC Symbol;Acc:2394] [Type: protein coding Ensembl/Havana merge]
Gene ID	ENSG00000108255
Location	17:27573481-27581512:1
Variations	Variation table
Source	e71

At the bottom of the page, there is a footer with 'Ensembl release 71 - April 2013 © WTSI / EBI' and links for 'About Ensembl', 'Privacy Policy', and 'Contact Us'. A 'Permanent link - View in archive site' is also present. An orange box highlights the 'ENSG00000108255' Gene ID, and a line connects it to a separate box labeled 'Ensembl ID'.

Ensembl ID

Indetificadores de Genes: HGNC

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

The screenshot displays the HGNC (HUGO Gene Nomenclature Committee) website. At the top left is the HGNC logo and the text 'HUGO Gene Nomenclature Committee'. To the right is a search bar labeled 'Search Genes'. Below the logo is a navigation menu with buttons for 'Home', 'Search Genes', 'Downloads', 'Gene Families', 'HCOP', 'Useful Links', 'About', and 'Contact Us'. A 'Request Symbol' button is located on the far right of the menu. The main content area features a grid of human chromosomes labeled 1 through 22, X, and Y. To the right of the chromosomes is a text block: '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](http://www.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.' Below this text is a 'Quick Gene Search' box with a search bar containing 'crystallin|beta A1', a search button, and options for search criteria (equal, begin, contain) and display hits (50).

Indetificadores de Genes: HGNC

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

The screenshot shows the HGNC (HUGO Gene Nomenclature Committee) website. At the top, there is a search bar with the text "Search Genes" and a magnifying glass icon. Below the search bar is a navigation menu with buttons for "Home", "Search Genes", "Downloads", "Gene Families", "HCOP", "Useful Links", "About", "Contact Us", and a yellow "Request Symbol" button. The main content area is titled "Quick Gene Search". It contains a search form with the text "Search symbols, keywords or IDs for:" and options for "Results that" (equal, begin, contain) and "Display" (50 hits). The search input field contains "crystallin beta A1". Below the search form, there are pagination links: "First Page", "Prev Page", "1", "2", "3", "4", "...", "69", "Next Page". The search results are displayed in a table with 4 columns: "Approved Symbol", "Approved Name", "Location", and "Best Match".

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

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

Indetificadores de Genes: HGNC

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

HGNC
HUGO Gene Nomenclature Committee

Search Genes

Home Search Genes Downloads Gene Families HCOP Useful Links About Contact Us Request Symbol

Gene Symbol Report

CRYBA1

Approved Symbol	CRYBA1
Approved Name	crvstallin, beta A1
HGNC ID	HGNC: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
HCOP	D	
TreeFam	D	

NUCLEOTIDE SEQUENCES

RefSeq:NM_005208	C	
CCDS:CCDS11249.1	C	
Vega:OTTHUMG00000132729	C	

GENE RESOURCES

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

PROTEIN RESOURCES

UniProtKB:P05813	D	
InterPro	D	
OMIM	D	
GeneTests	D	
Orphanet	D	

CLINICAL RESOURCES

DECIPHER	D	
COSMIC	D	
LSDB:LOVD - Leiden Open Variation Database	C	
Genetic Testing Registry	C	

Symbol

HGNC ID

Más Identificadores

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

Indetificadores 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

[Sample Searches](#)
[OMIM Tutorial](#)

Advanced Search: [OMIM](#), [Clinical Synopses](#), [OMIM Gene Map](#) , [Search History](#)



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

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

Indetificadores 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 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 NCBI)

Gene Phenotype Relationships

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

External Links:

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

OMIM ID

*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 NCBI)

Gene Phenotype Relationships

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

Symbol

HGNC Approved Gene Symbol: CRYBA1

Identificadores de Genes: Orphanum

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

The screenshot shows the Orphanet website homepage. At the top, there is a navigation bar with the Orphanet logo, the tagline "The portal for rare diseases and orphan drugs", and logos for Inserm, France, and the European Union. A language selector shows "EN" (English) is selected. A search bar is present with an "OK" button. Below the search bar, there are several service categories: "Access our Services" (highlighted with an orange box), "Read Orphanet reports" (green box), and "Contribute to Orphanet" (green box). The "Access our Services" section lists various tools and directories, with a red box highlighting the "Inventory, classification and encyclopaedia of rare diseases, with genes involved" and an arrow pointing to a search box. The "Read Orphanet reports" section lists various reports and surveys. The "Contribute to Orphanet" section includes options to register activity and sponsor Orphanet. Below these are sections for "Download Orphanet data", "Newsletter", "Other documents", "Other rare diseases websites", and "Events". The "Events" section shows an event for "Myasthenia 2013" on July 1-2, 2013, in Paris, France.

Búsquedas de Genes y Enfermedades

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

Indetificadores de Genes: Orphanum

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

The screenshot shows the Orphanet website interface. At the top, there are language options (FR, EN, ES, DE, IT, PT) and a navigation menu with categories like 'Rare diseases', 'Orphan drugs', 'Expert centres', 'Diagnostic tests', 'Research and trials', 'Patient organisations', 'Professionals and institutions', and 'Other information'. The 'Genes' category is highlighted. Below the navigation, there is a search bar with the text 'beta crystallin a1' and a dropdown menu for search criteria: 'Gene name or symbol' (selected), 'MIM number (Gene)', 'Disease name', and 'MIM number (disease)'. The search results show the gene 'CRYBA1 - Crystallin, beta A1' with various identifiers: Orpha number (ORPHA120836), OMIM (123610), HGNC (2394), UniProtKB (P05813), Genatlas (CRYBA1), Ensembl (ENSG00000108255), IUPHAR-DB (-), and Reactome (-).

Indicar tipo de búsqueda: Gen

Formulario de búsqueda: Indicar tipo de identificador

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

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

Identificadores de Enfermedades: OMIM

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

The screenshot shows the OMIM website interface. At the top, there is a navigation menu with links: Home, About, Statistics, Downloads/API, Help, External Links, Terms of Use, and Contact Us. Below the menu is a search bar containing the text "friedreich ataxia". To the right of the search bar is a "Search" button and a "Sort by:" dropdown menu with "Relevance" selected and "Date updated" as an option. Below the search bar, there are links for "Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map" and "Toggle: search terms highlighted". A "Search History: View, Clear" link is also present. The search results are displayed in a light blue box. At the top of the results is the identifier "#229300" in a rounded rectangle. Below it is the title "FRIEDREICH ATAXIA 1; FRDA". Underneath, there is a section for "Alternative titles; symbols" listing "FRDA1" and "FA". A section titled "Other entities represented in this entry:" lists "FRIEDREICH ATAXIA WITH RETAINED REFLEXES, INCLUDED; FARR, INCLUDED". At the bottom, there is a section for "Phenotype Gene Relationships" which contains a table with the following data:

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: Orphanum

- Enfermedades Raras

The screenshot shows the Orphanet website interface. At the top, there is a navigation bar with the Orphanet logo, the text "The portal for rare diseases and orphan drugs", and logos for Inserm, France, and the European Union. A language selector shows "EN" selected. A navigation menu includes categories like "Rare diseases", "Orphan drugs", "Expert centres", etc. The main content area shows a search for "Friedreich ataxia" with a dropdown menu for search options: "Disease name" (selected), "Gene name or symbol", "OMIM", "ICD-10", and "Orpha number". Below the search results, a table lists various identifiers for Friedreich ataxia.

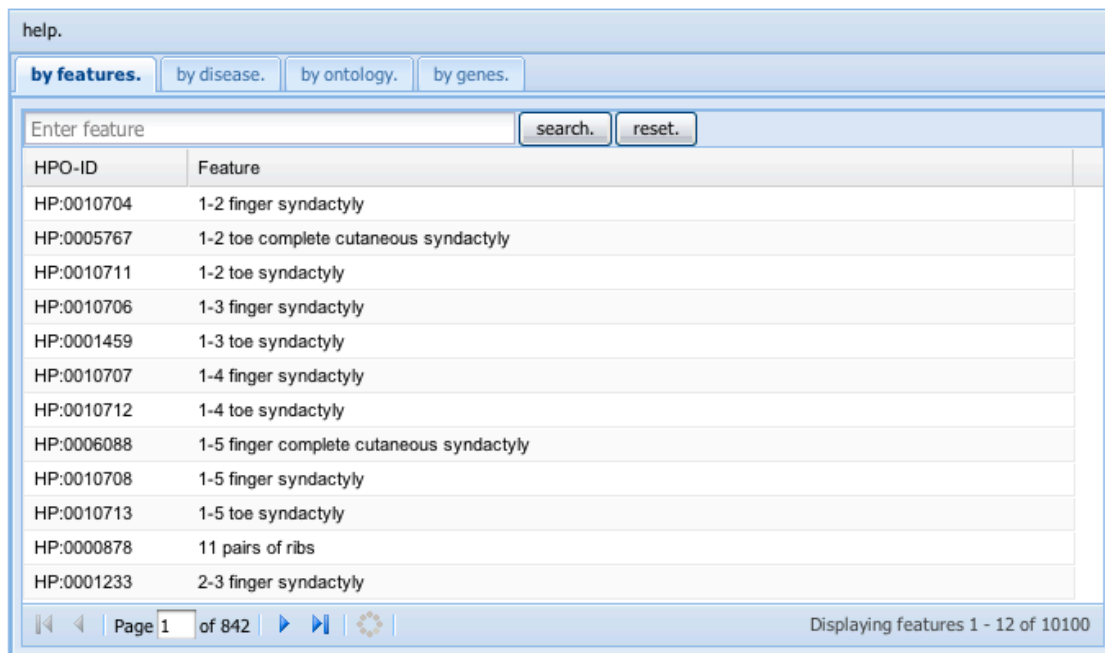
Orpha number	: ORPHA95	ICD-10	: G11.1
Synonym(s)	: -	OMIM	: 229300 601992
Prevalence	: 1-9 / 100 000	UMLS	: C0016719
Inheritance	: Autosomal recessive	MeSH	: D005621
Age of onset	: Childhood	MedDRA	: 10017374
		SNOMED CT	: 10394003

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

Fenotipos

- PhenExplorer

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



help.

by features. by disease. by ontology. by genes.

Enter feature search. reset.

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

Page 1 of 842 | Displaying features 1 - 12 of 10100

Entrada a PhenUMA

1. Opciones de Entrada

3. Opciones de la Red de Salida

The screenshot shows the PhenUMA web interface. At the top, there is a navigation bar with links for 'Main', 'Tutorial', 'How to cite us', and 'Contact us'. Below this is the PhenUMA logo and the title 'PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases'. The main form contains several input fields and a button. A red box highlights the 'Type of Input' and 'ID' dropdowns, with a red arrow pointing to the '1. Opciones de Entrada' label. A green box highlights the 'Type of Output Network' and 'Confidence' dropdowns, with a green arrow pointing to the '3. Opciones de la Red de Salida' label. A blue box highlights the 'Input List' text area, with a blue arrow pointing to the '2. Datos de Entrada' label. A 'Load From File' link is located to the right of the input list, and a 'Build Network' button is at the bottom center.

Main Tutorial How to cite us Contact us

PhenUMA

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:

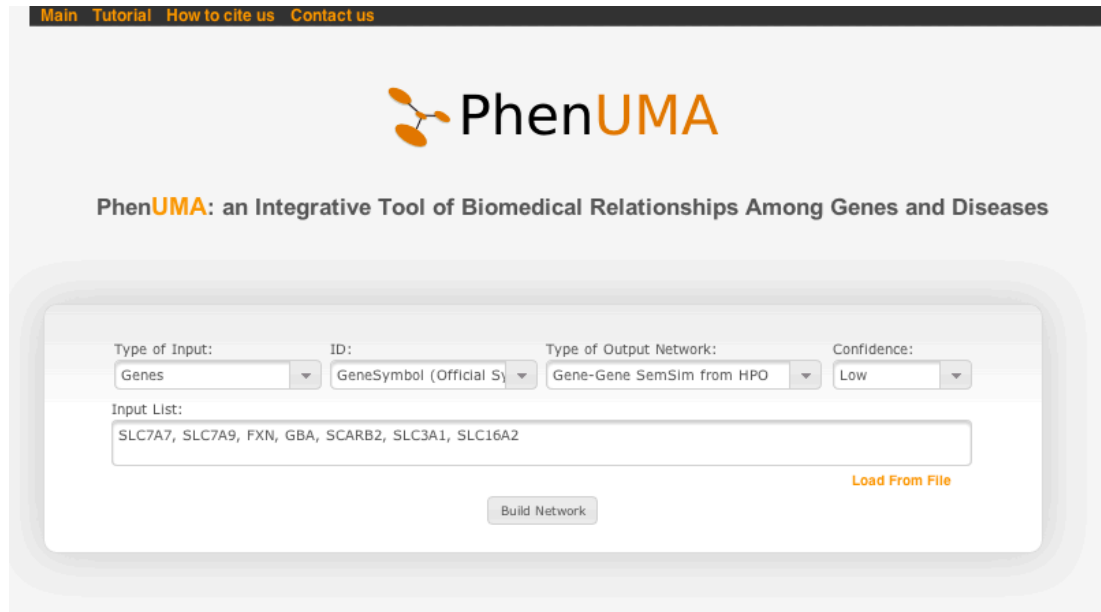
Load From File

Build Network

2. Datos de Entrada

2. Datos de Entrada

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

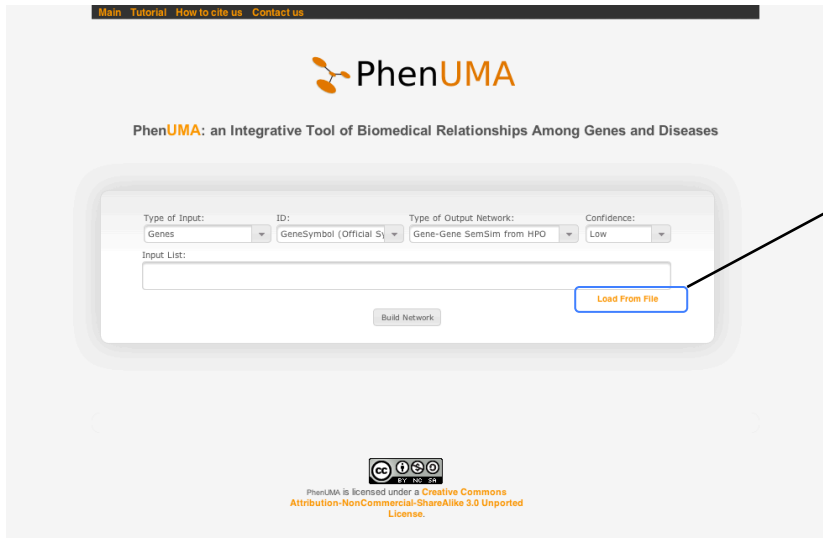


The screenshot shows the PhenUMA web interface. At the top, there is a navigation bar with links for "Main", "Tutorial", "How to cite us", and "Contact us". Below this is the PhenUMA logo, which consists of three orange circles connected by lines, followed by the text "PhenUMA". Underneath the logo is the tagline "PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases". The main content area contains a form with the following fields:

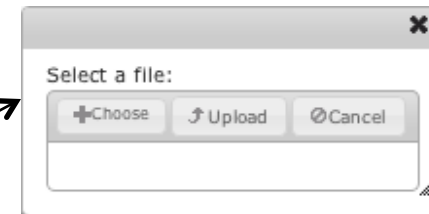
- Type of Input:** A dropdown menu set to "Genes".
- ID:** A dropdown menu set to "GeneSymbol (Official Sy".
- Type of Output Network:** A dropdown menu set to "Gene-Gene SemSim from HPO".
- Confidence:** A dropdown menu set to "Low".
- Input List:** A text input field containing the text "SLC7A7, SLC7A9, FXN, GBA, SCARB2, SLC3A1, SLC16A2".
- Buttons:** A "Build Network" button and a "Load From File" link.

2. Datos de Entrada

- Cargar entrada desde fichero de texto



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

1. Opciones de Entrada

3. Opciones de la Red de Salida

The screenshot shows the PhenUMA web interface. At the top, there is a navigation bar with links for 'Main', 'Tutorial', 'How to cite us', and 'Contact us'. Below this is the PhenUMA logo and the title 'PhenUMA: an Integrative Tool of Biomedical Relationships Among Genes and Diseases'. The main form contains several input fields and a button. A red box highlights the 'Type of Input' and 'ID' dropdowns, with a red arrow pointing to the 'Type of Input' dropdown. A green box highlights the 'Type of Output Network' and 'Confidence' dropdowns, with a green arrow pointing to the 'Type of Output Network' dropdown. A blue box highlights the 'Input List' text area, with a blue arrow pointing to it. A 'Load From File' link is located to the right of the 'Input List' field. A 'Build Network' button is at the bottom of the form.

Main Tutorial How to cite us Contact us

PhenUMA

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:

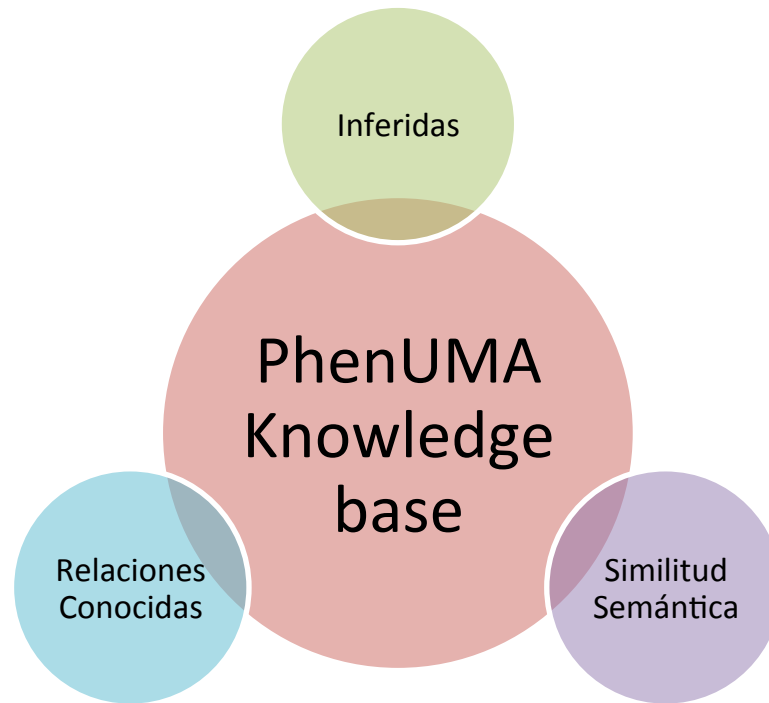
Load From File

Build Network

2. Datos de Entrada

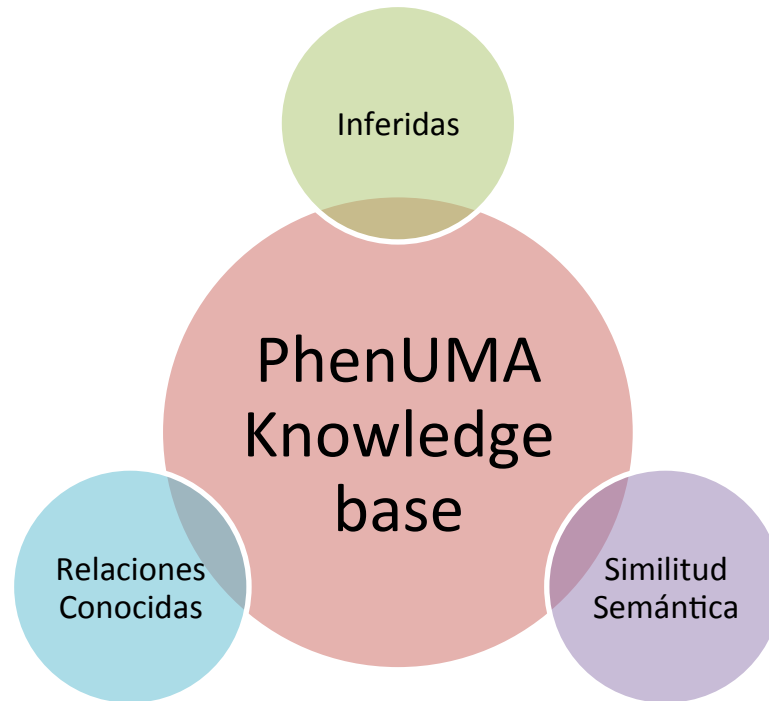
3. Opciones de Salida: Knowledge Base

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



Knowledge Base

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



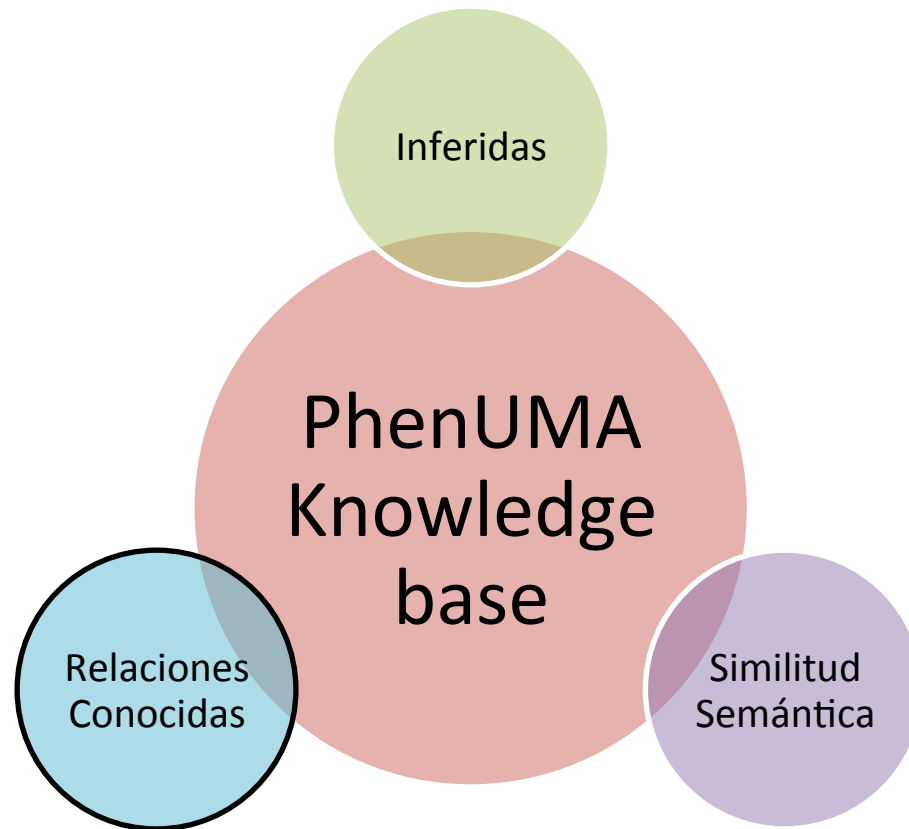
Gene-Gene Known Protein-Protein Interaction from STRING
Gene-Gene Known Metabolic Interactions (Veeramani et al.)

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)

Entrada a PhenUMA → 3.Opciones de la Red de Salida → Knowledge Base

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.

OMIM[®]

Online Mendelian Inheritance in Man[®]

An Online Catalog of Human Genes and Genetic Disorders

Updated 7 June 2013

orphanet

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.

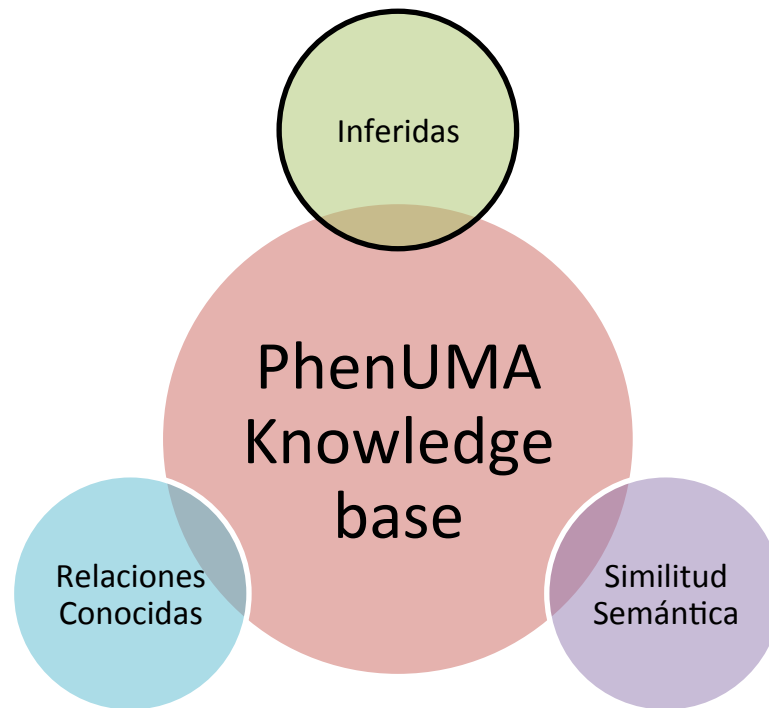
Description

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

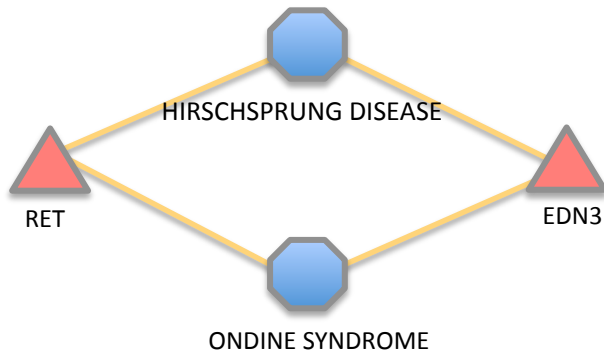


Gene-Gene Inferred from Orphanet

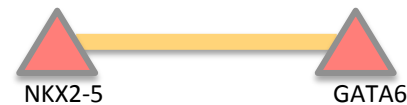
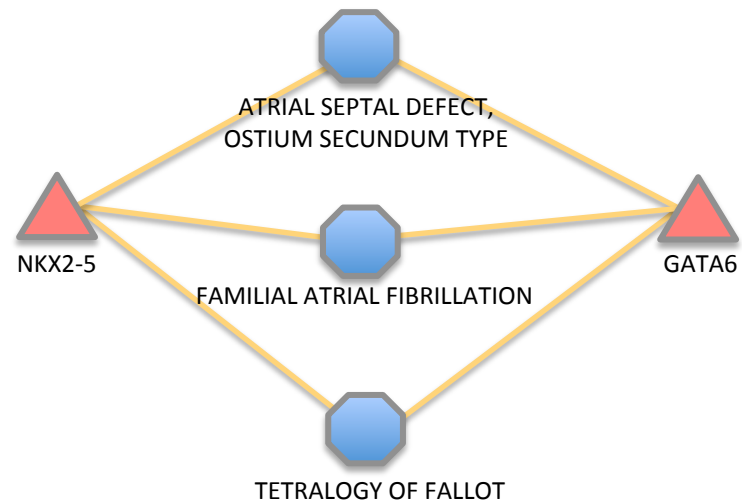
Orpha-Orpha Inferred from Gene

Knowledge Base : Relaciones Inferidas

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

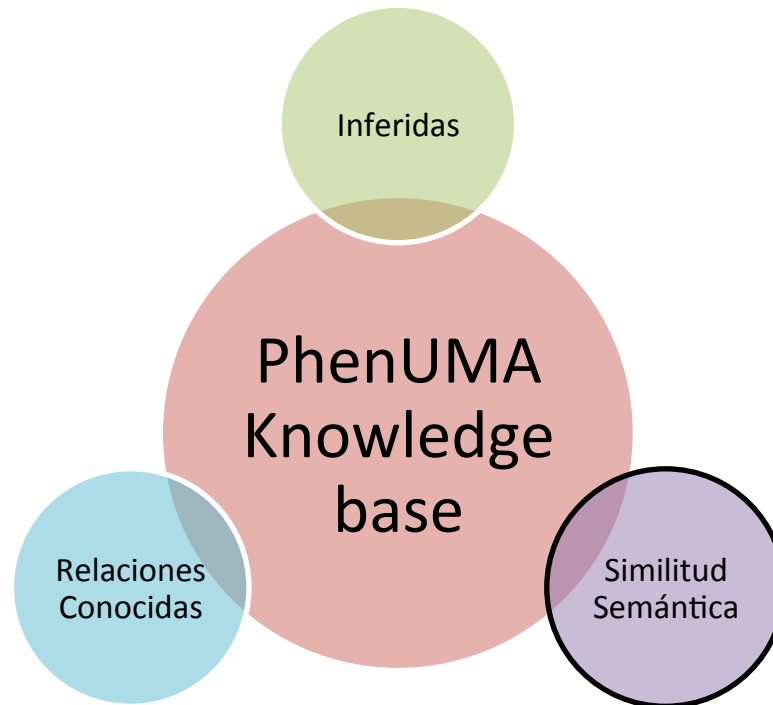


Gene-Gene Inferred from Orphanet



Orpha-Orpha Inferred from Gene

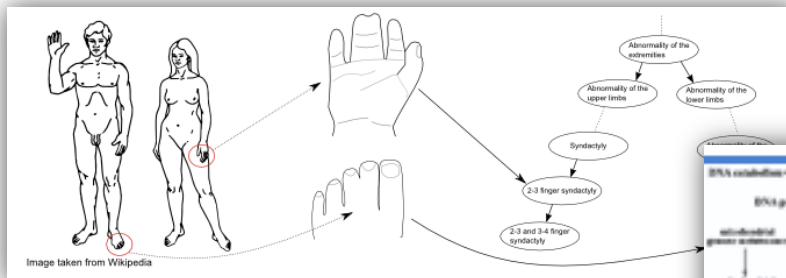
Knowledge Base



Entrada a PhenUMA → 3.Opciones de la Red de Salida → 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**

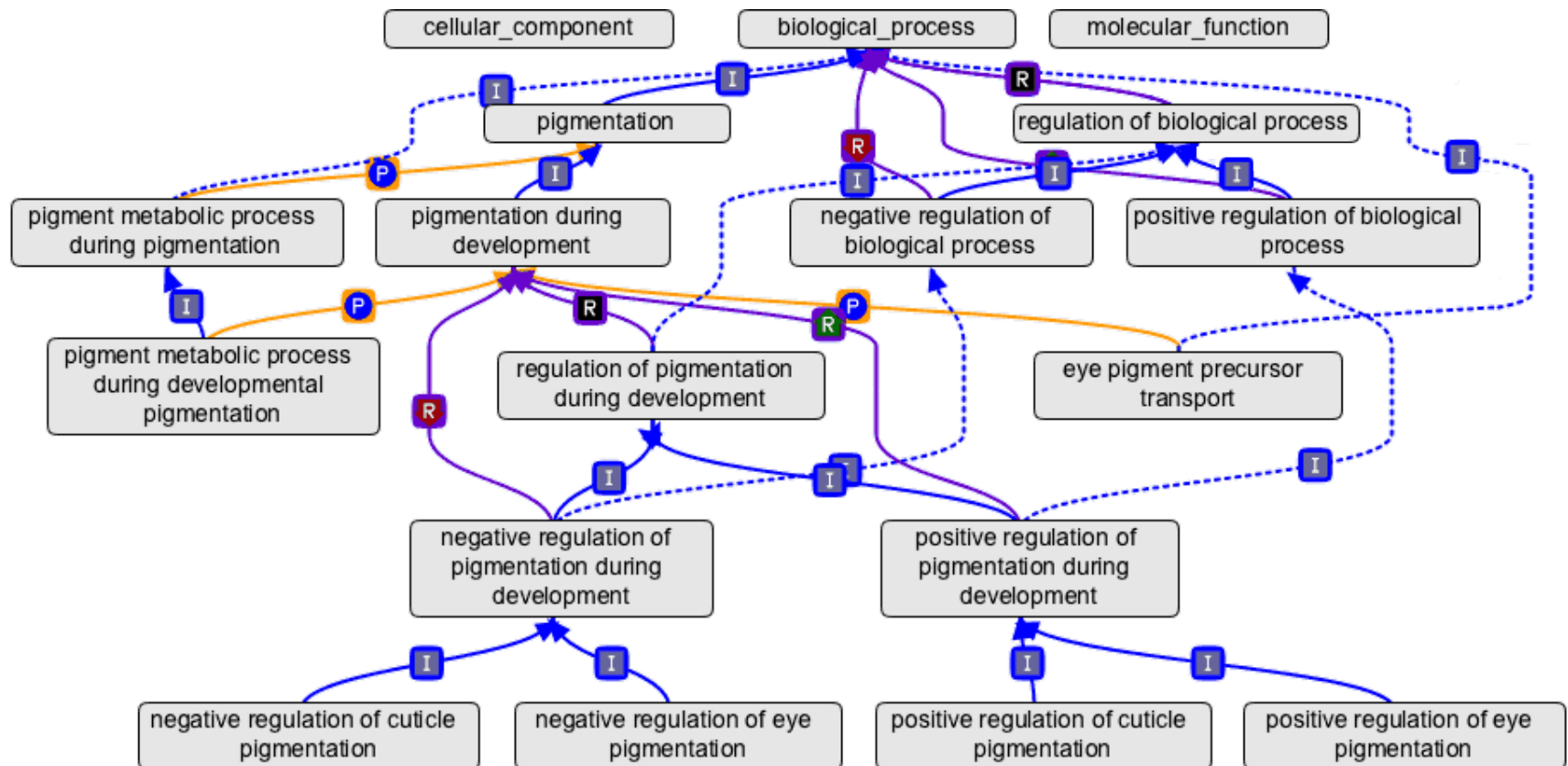


Human Phenotype Ontology

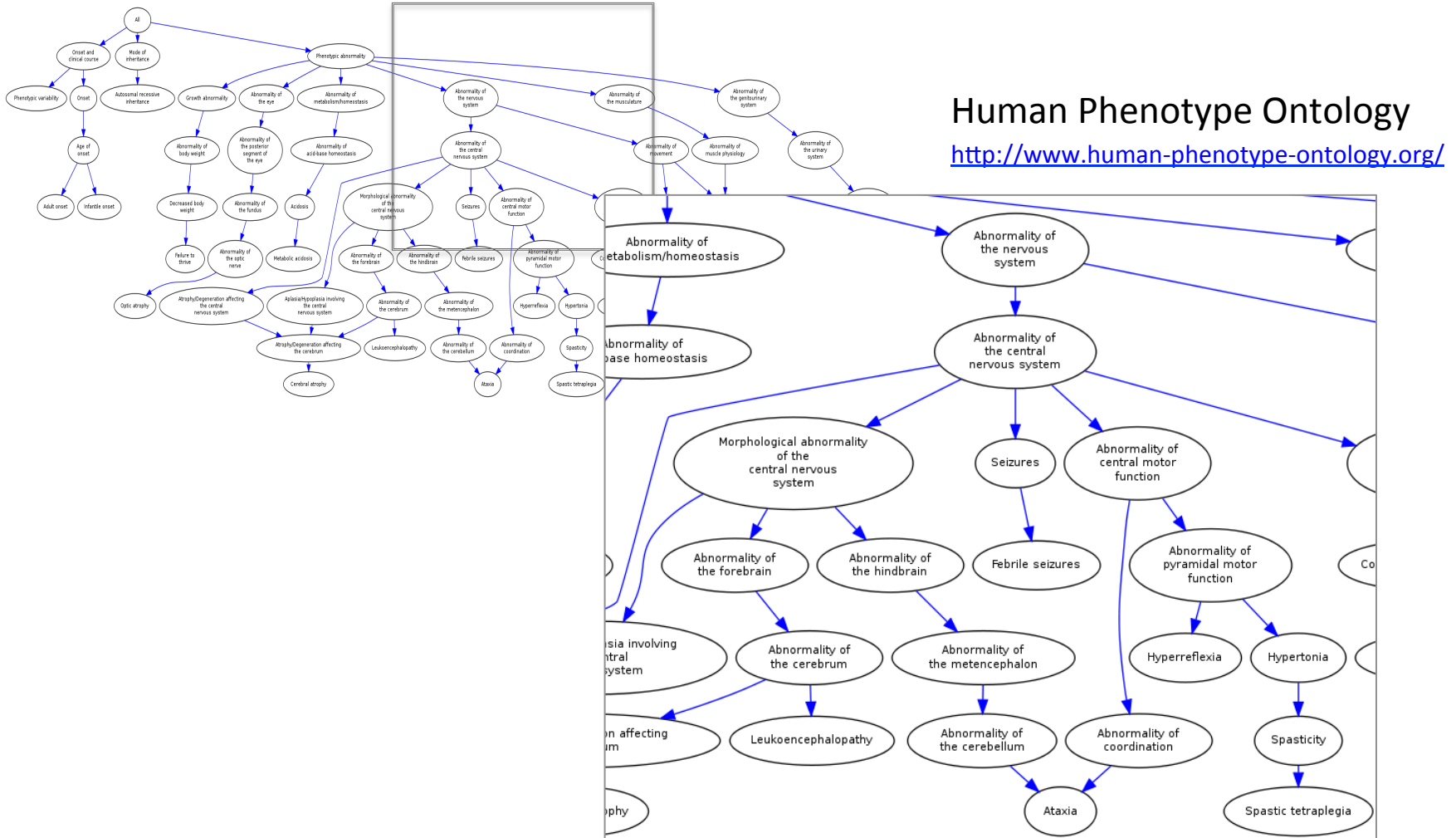


Knowledge Base : Similitud Semántica

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



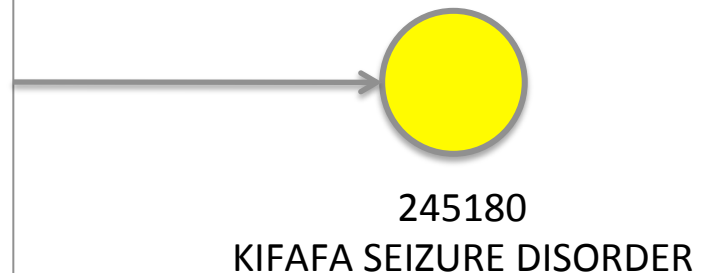
Knowledge Base : Similitud Semántica



Knowledge Base : Similitud Semántica

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

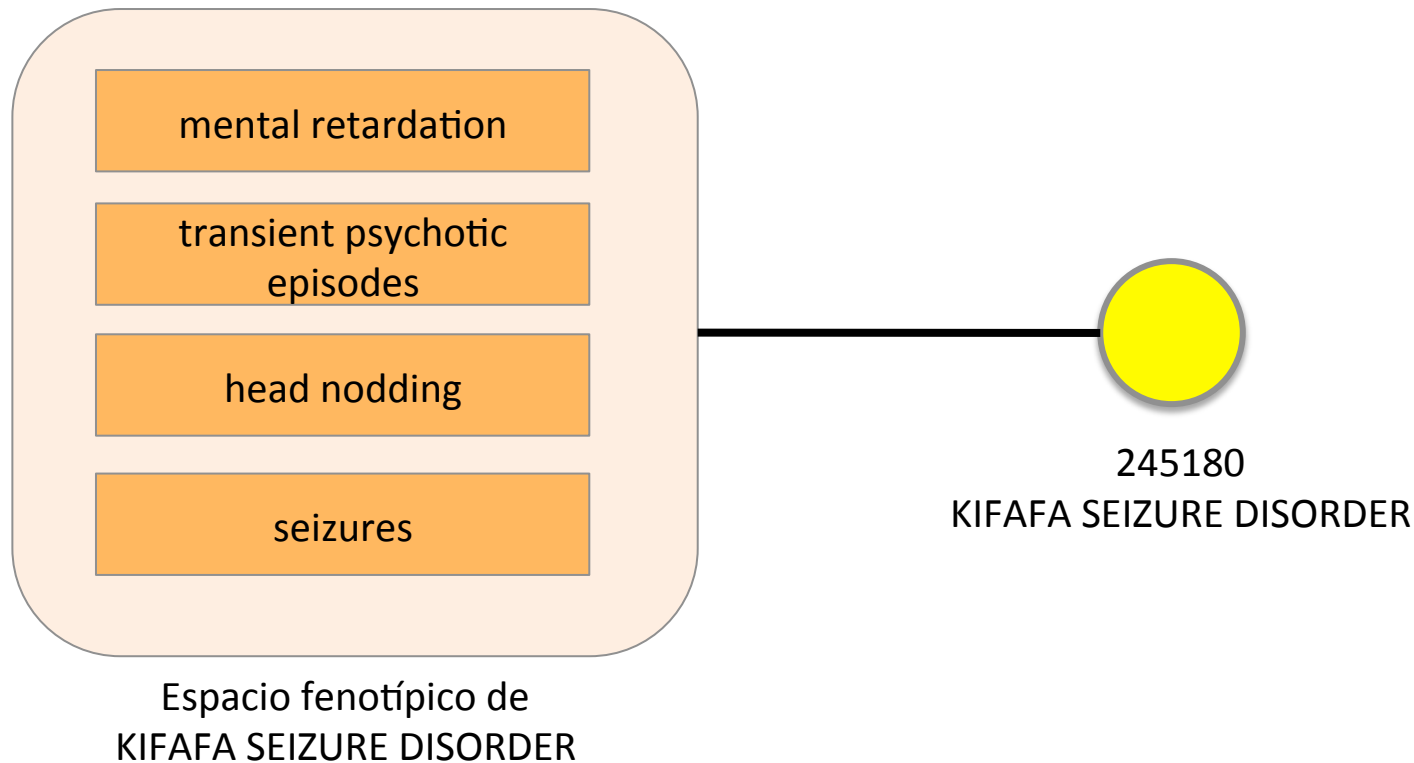
... Many showed parkinsonian features and-or other neurologic abnormalities, as well as **mental retardation** and **transient psychotic episodes**. In children, **head nodding** was a frequent precursor of later grand mal **seizures**...



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

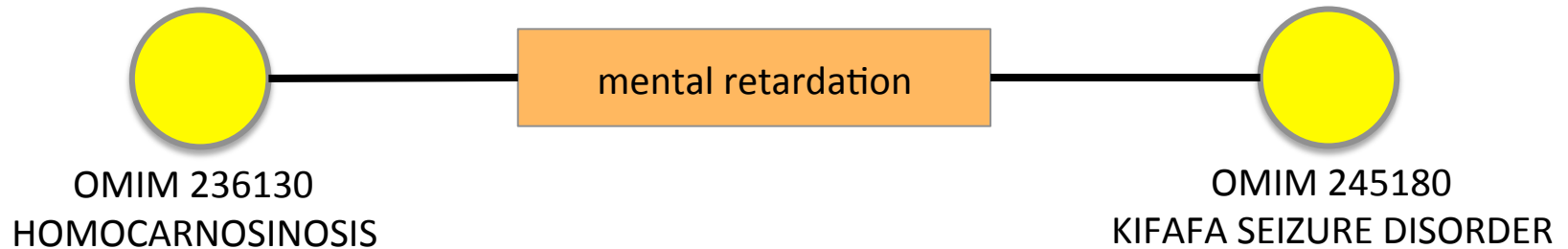
Knowledge Base : Similitud Semántica

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



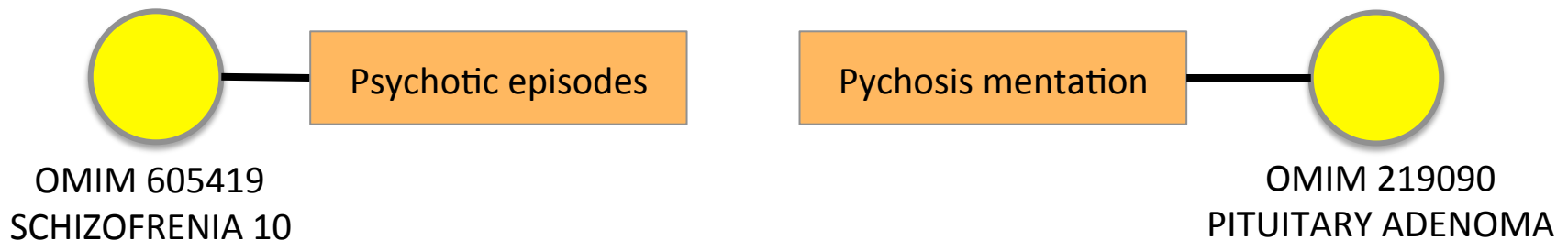
Knowledge Base : Similitud Semántica

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



Knowledge Base : Similitud Semántica

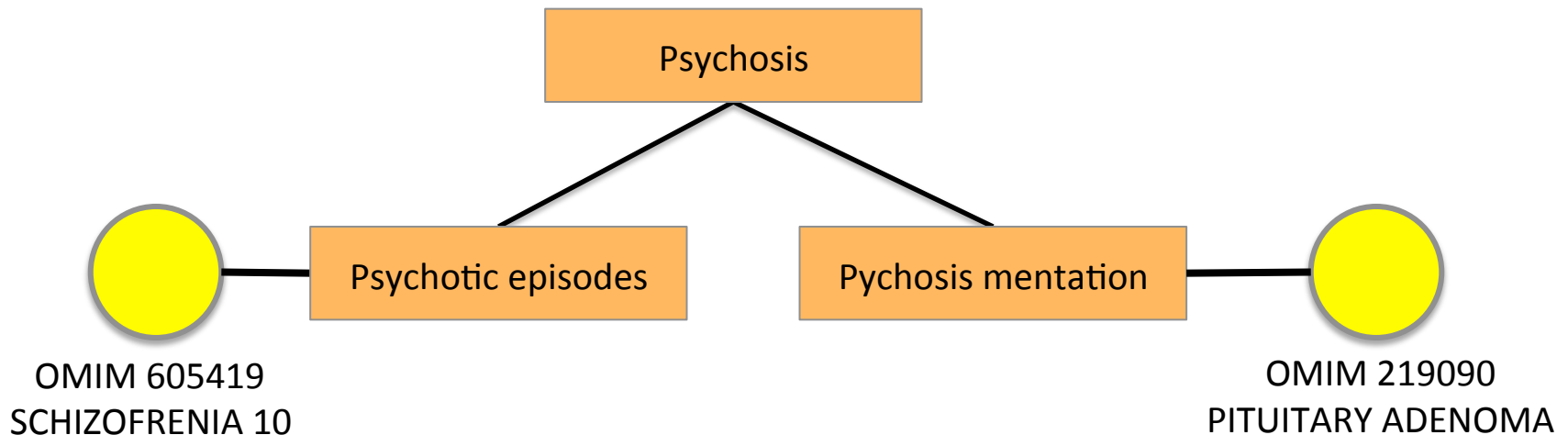
- La estructura jerárquica de la ontología permite ampliar las relaciones que pueden establecerse a partir de inferencias.



¿Deberían estar relacionadas?

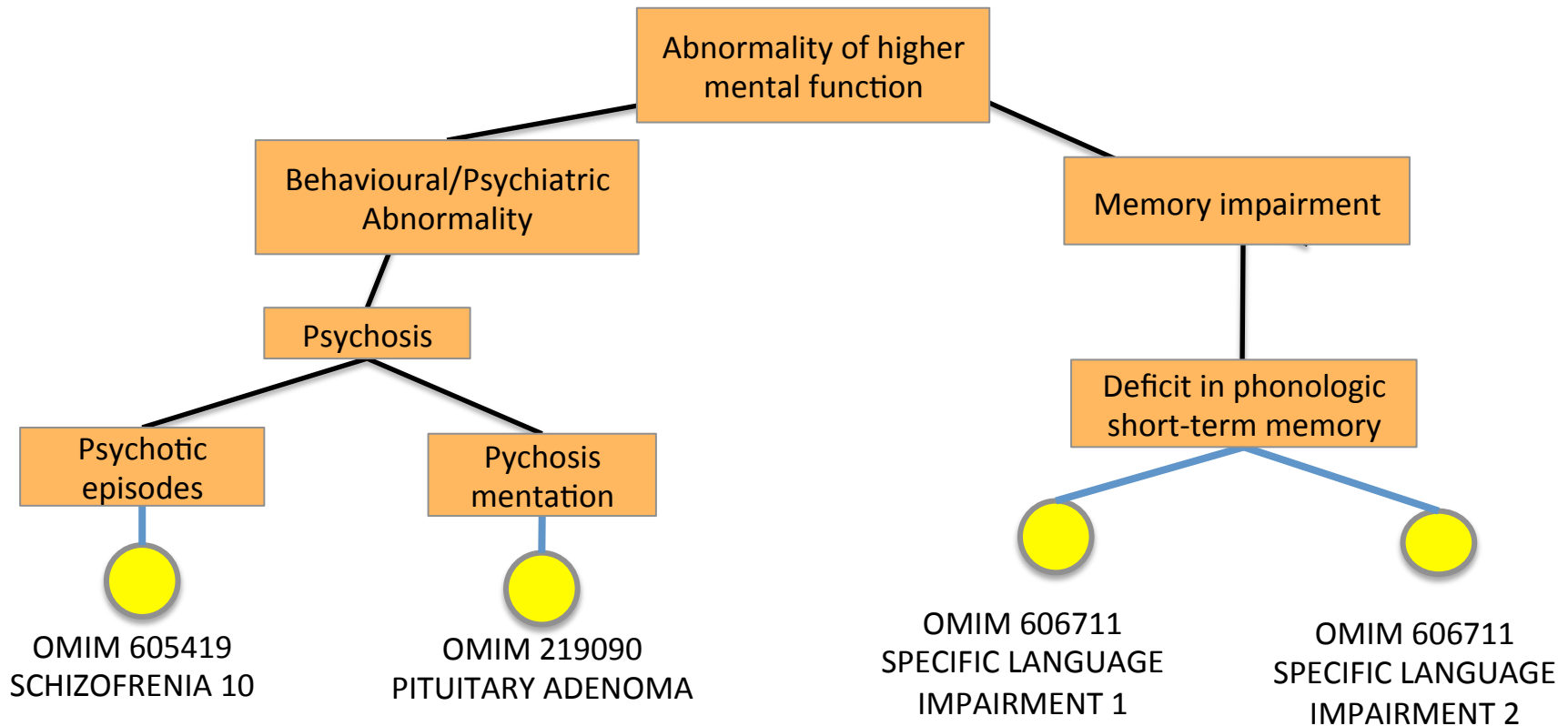
Knowledge Base : Similitud Semántica

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



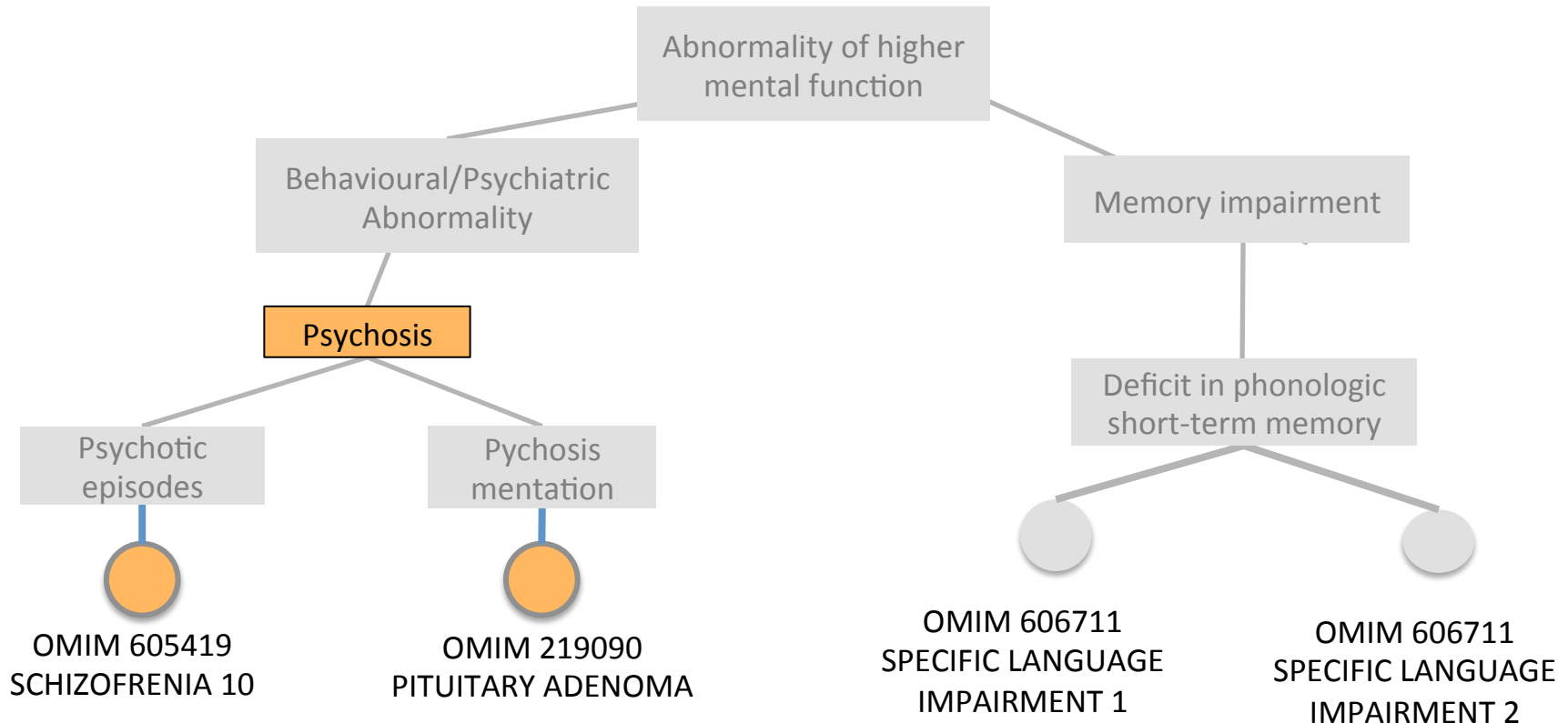
Knowledge Base : Similitud Semántica

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



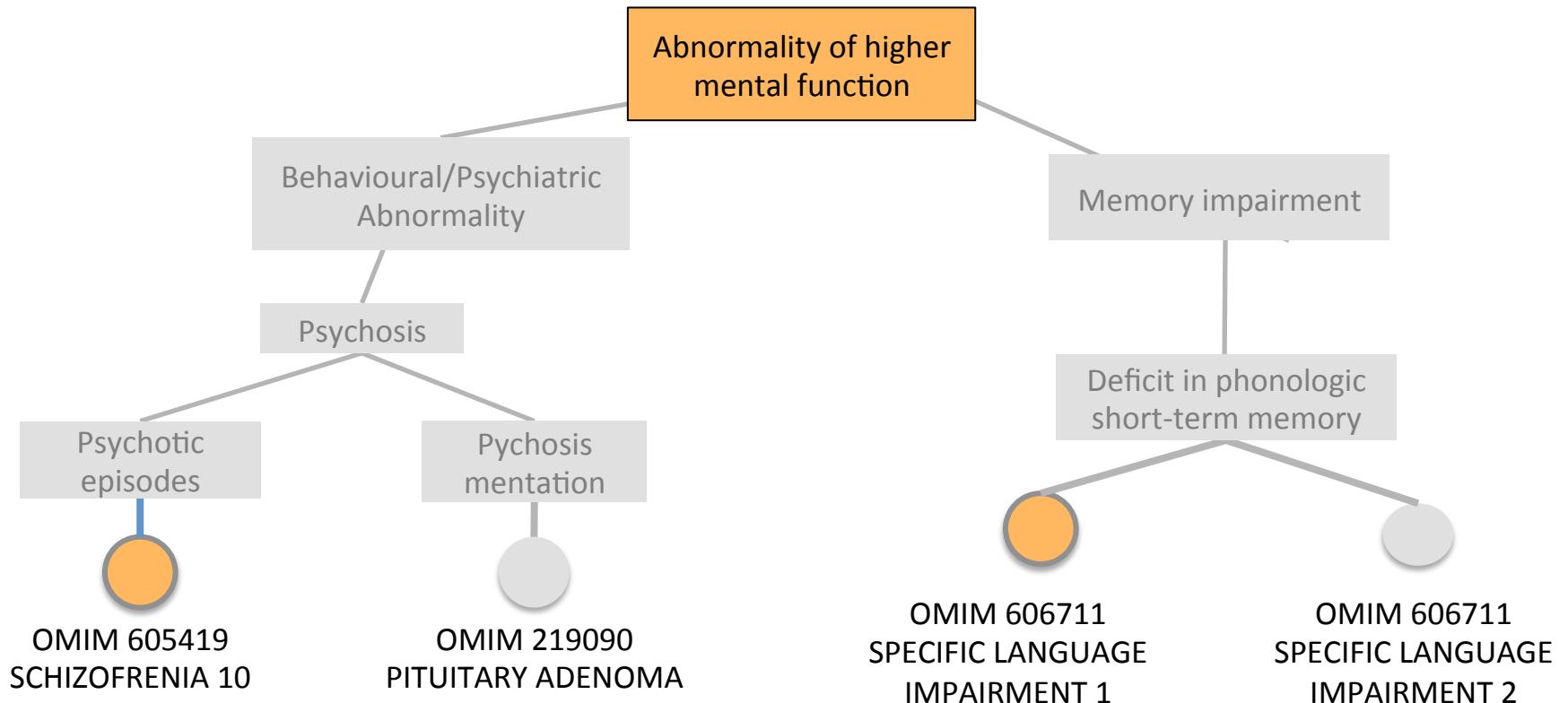
Knowledge Base : Similitud Semántica

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



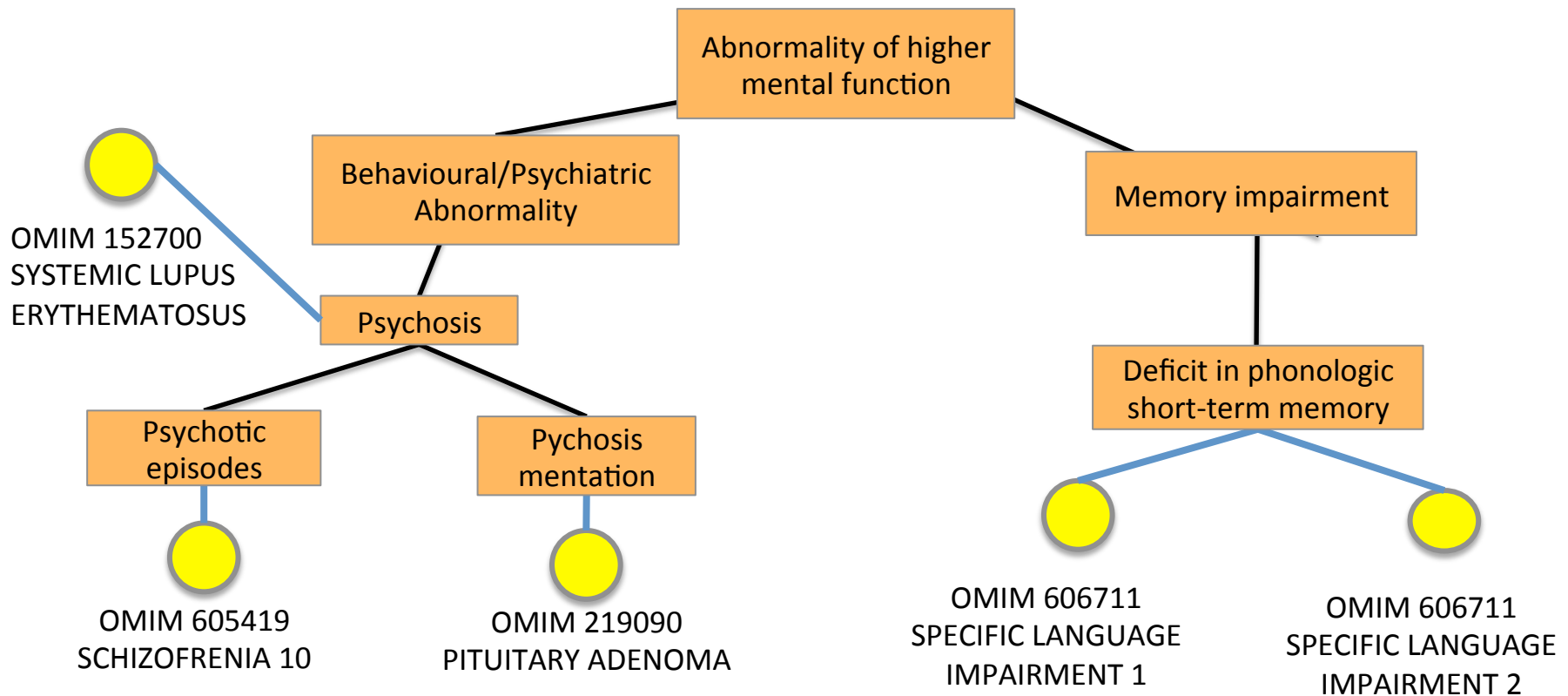
Knowledge Base : Similitud Semántica

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



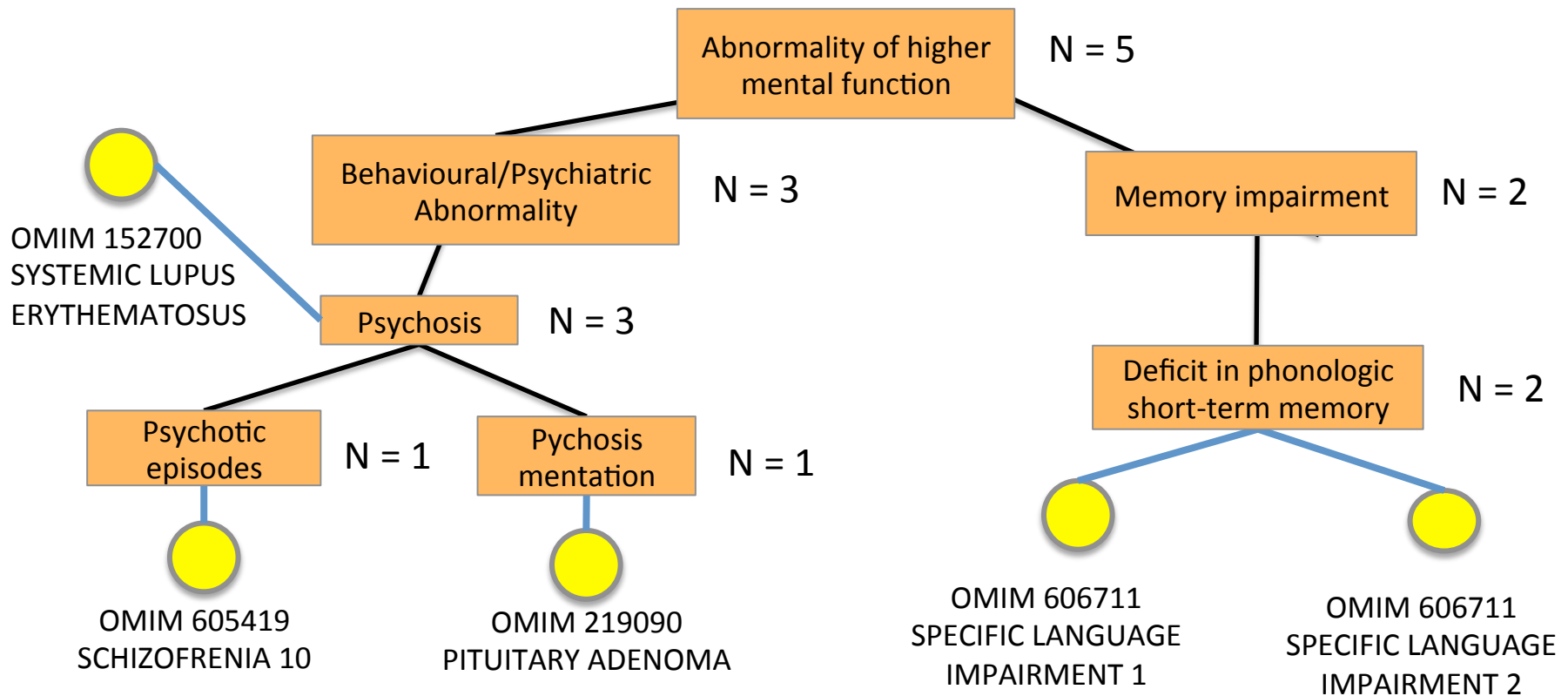
Knowledge Base : Similitud Semántica

- 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



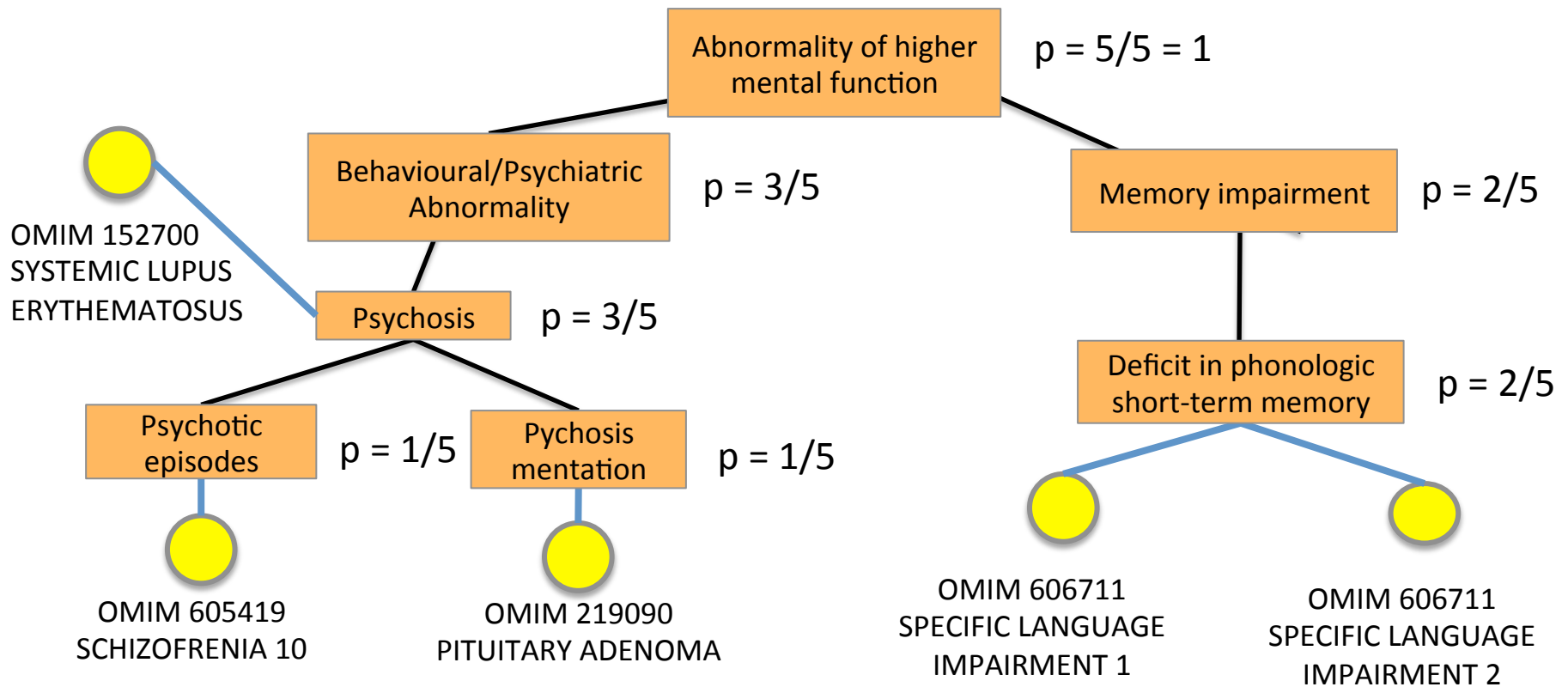
Knowledge Base : Similitud Semántica

- N = número de enfermedades asociadas a un fenotipo



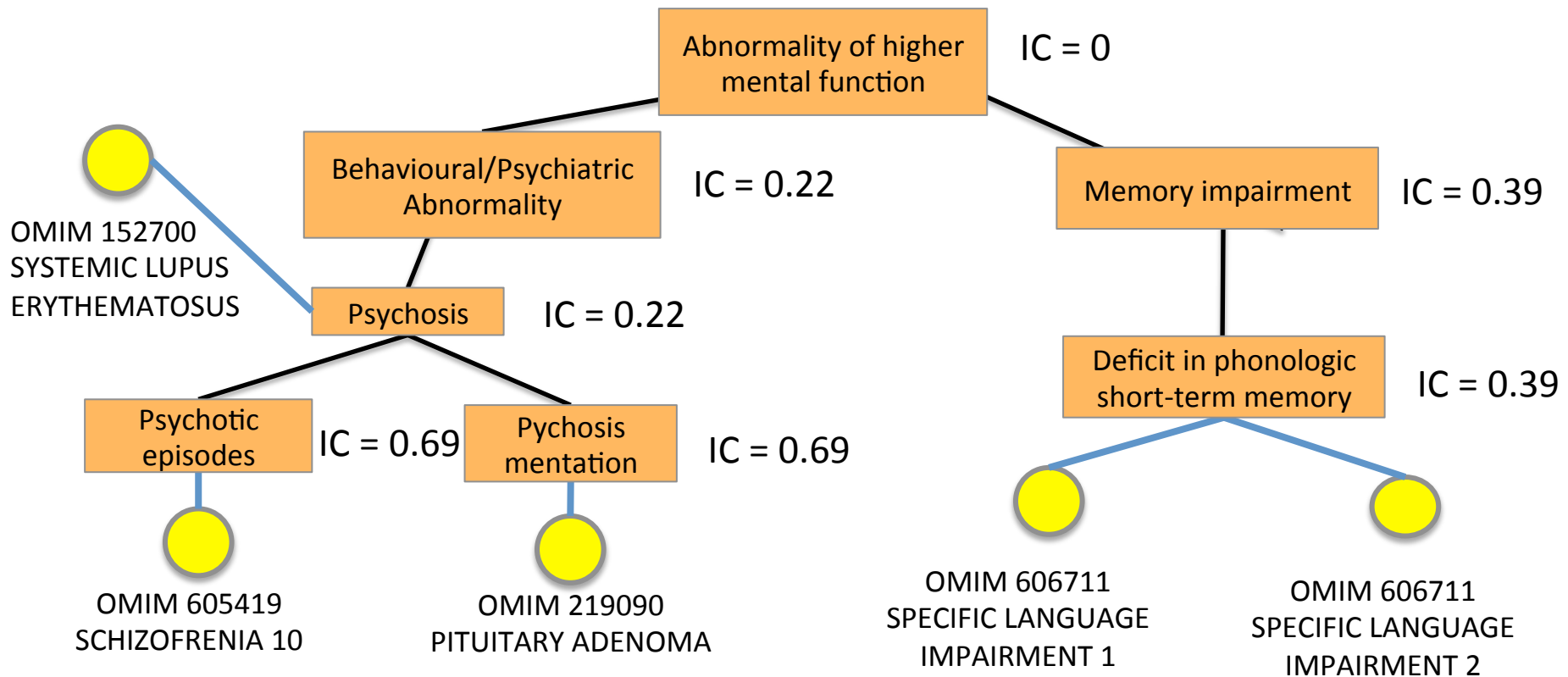
Knowledge Base : Similitud Semántica

- Probabilidad de cada término $p(t)$



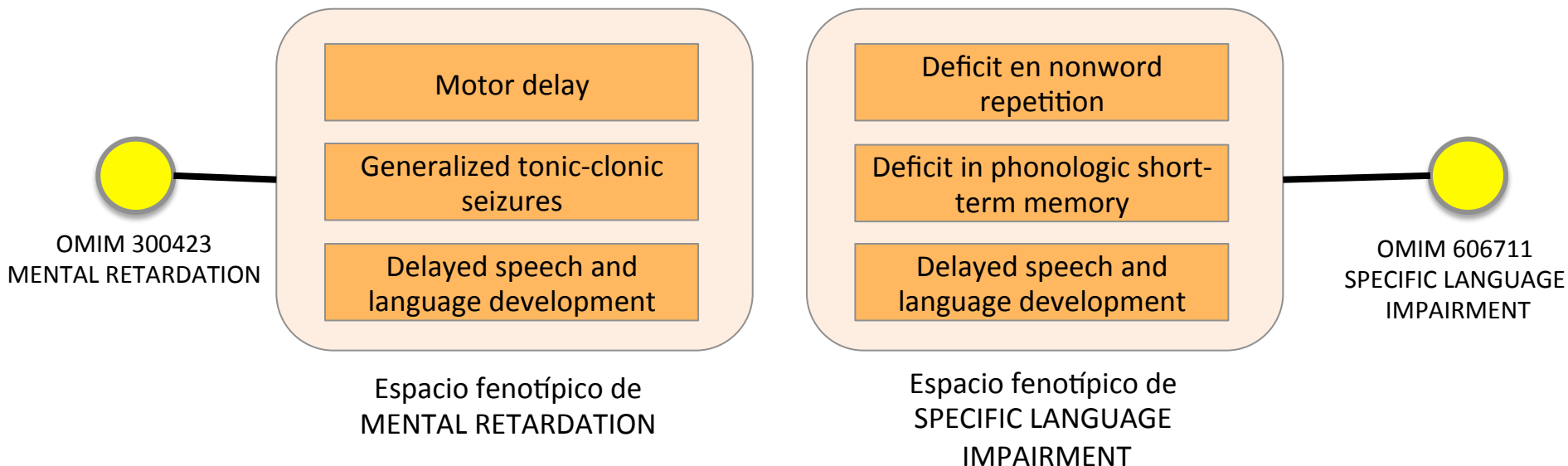
Knowledge Base: Similitud Semántica

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



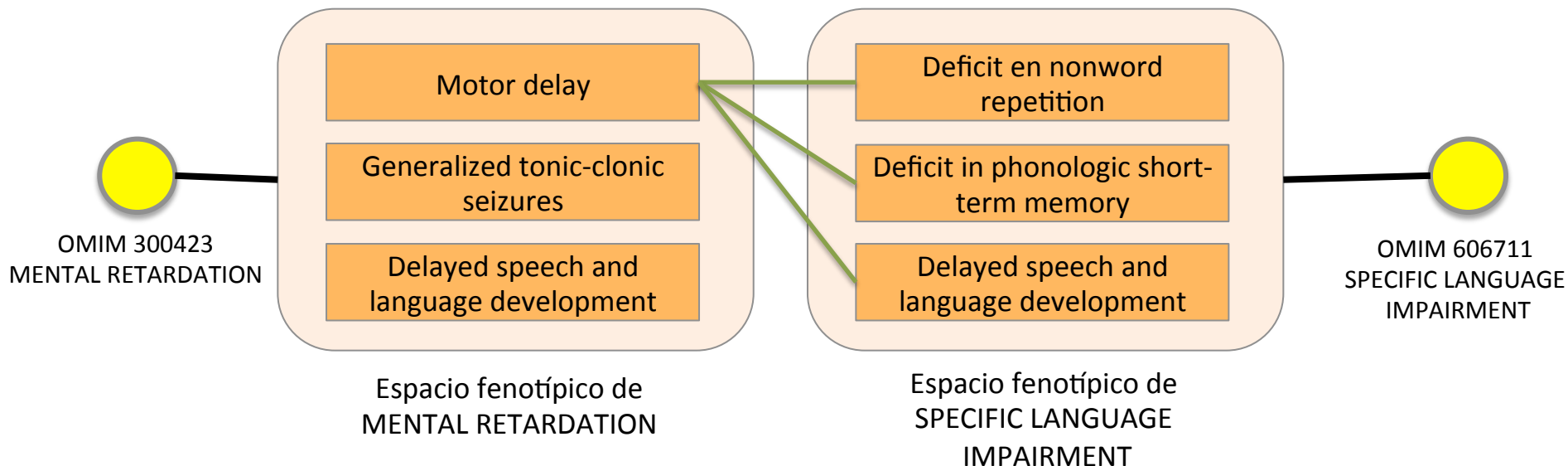
Knowledge Base : Similitud Semántica

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



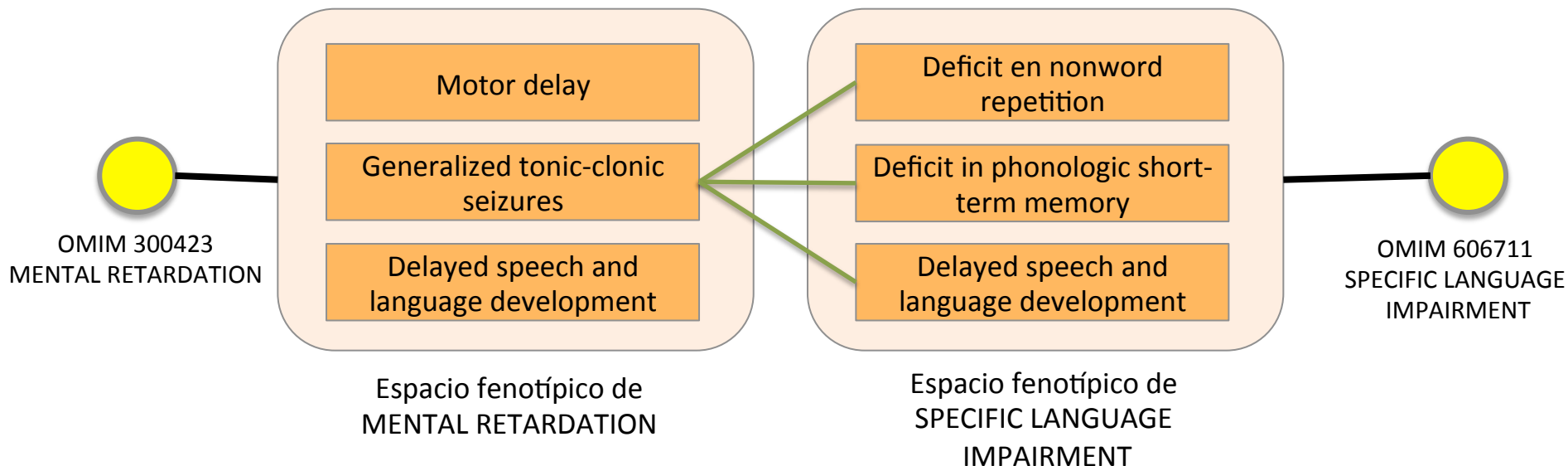
Knowledge Base : Similitud Semántica

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



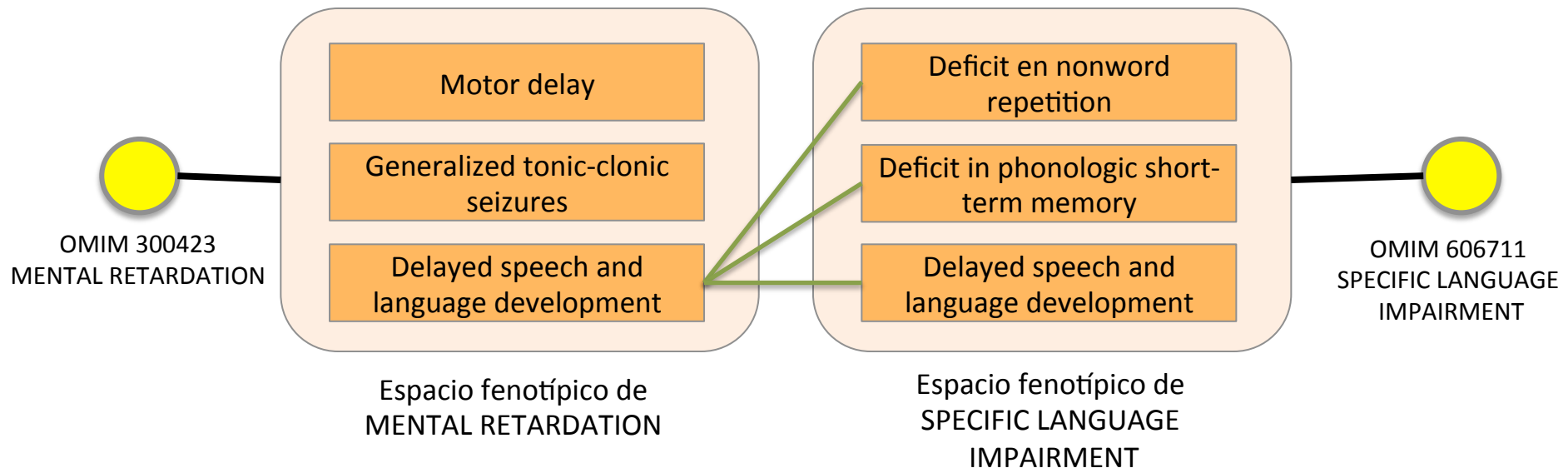
Knowledge Base : Similitud Semántica

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



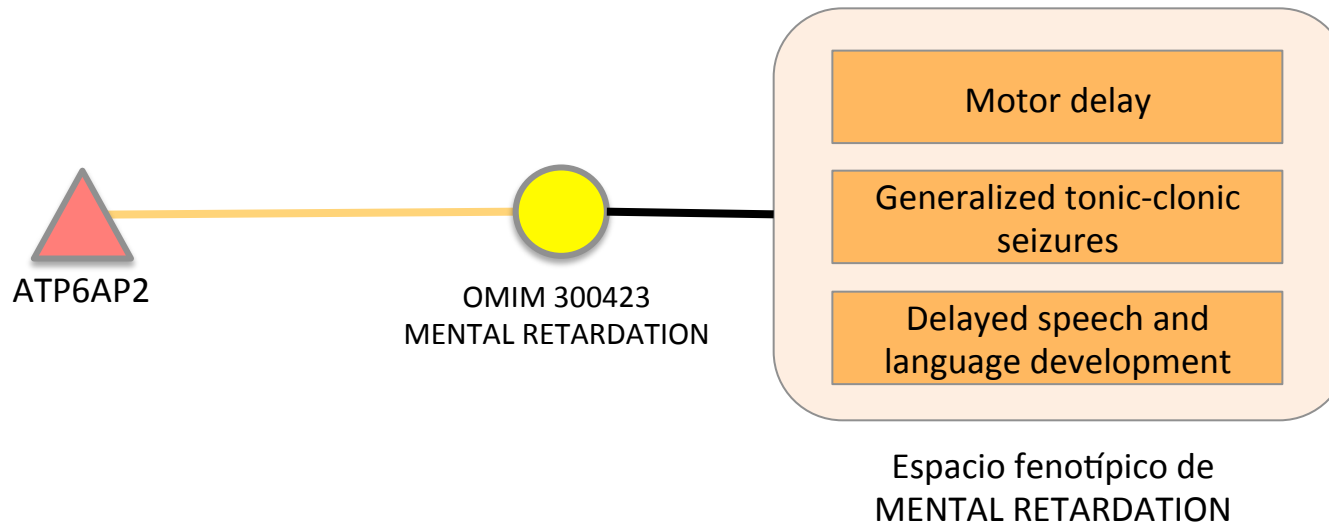
Knowledge Base : Similitud Semántica

- 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**



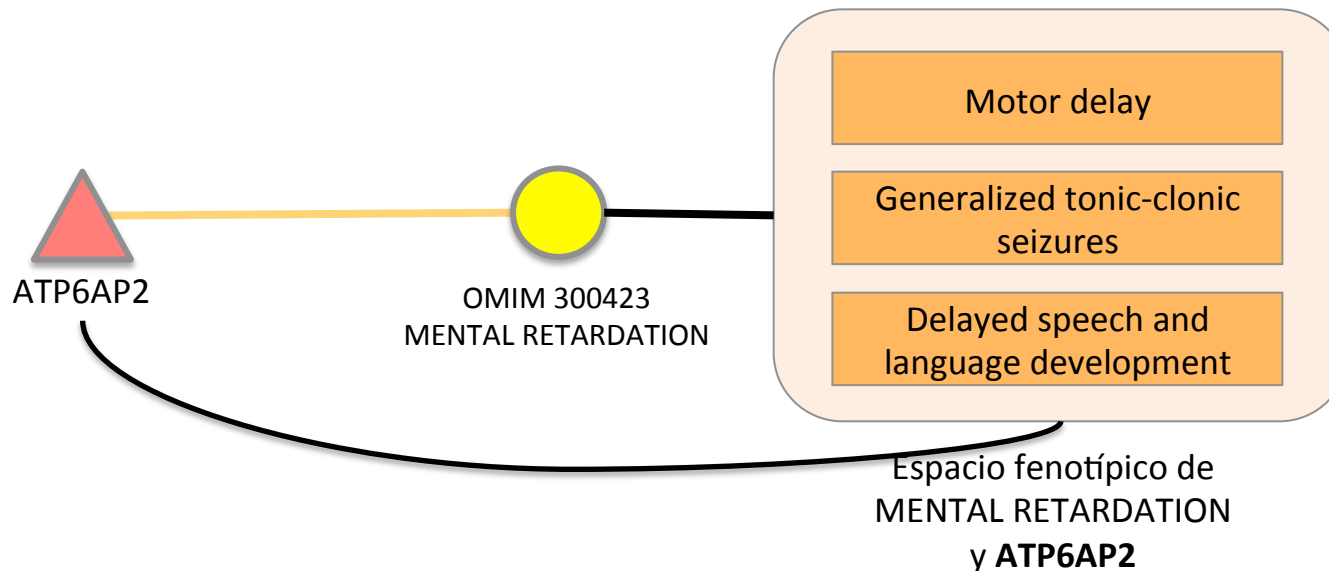
Knowledge Base : Similitud Semántica

- 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.



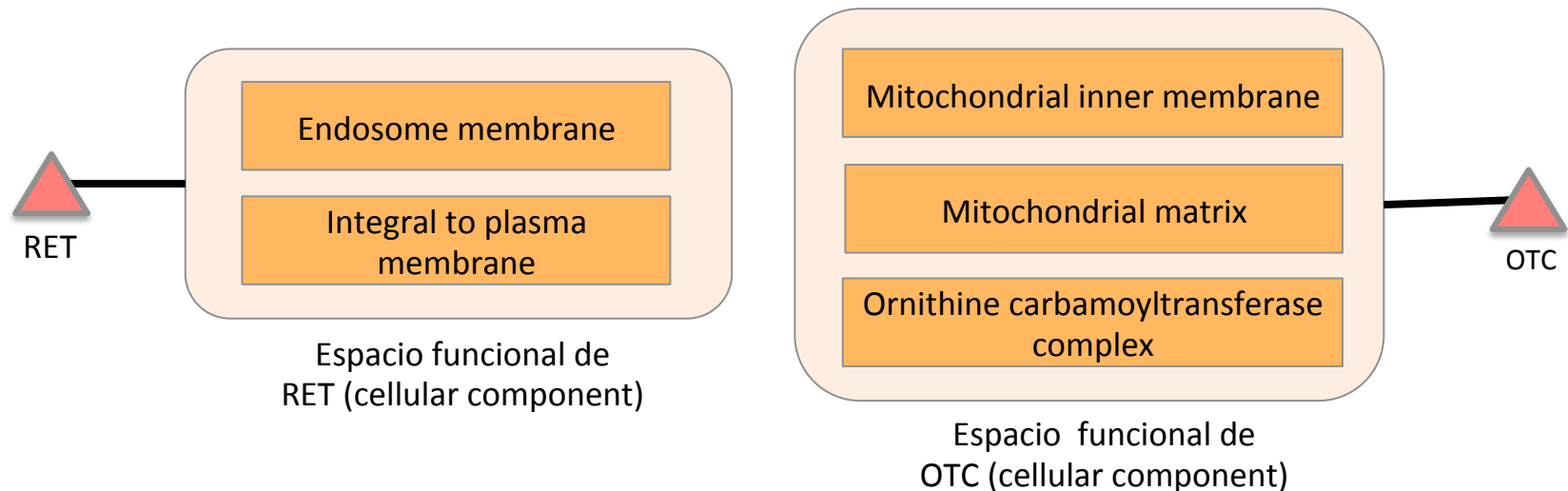
Knowledge Base : Similitud Semántica

- 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.



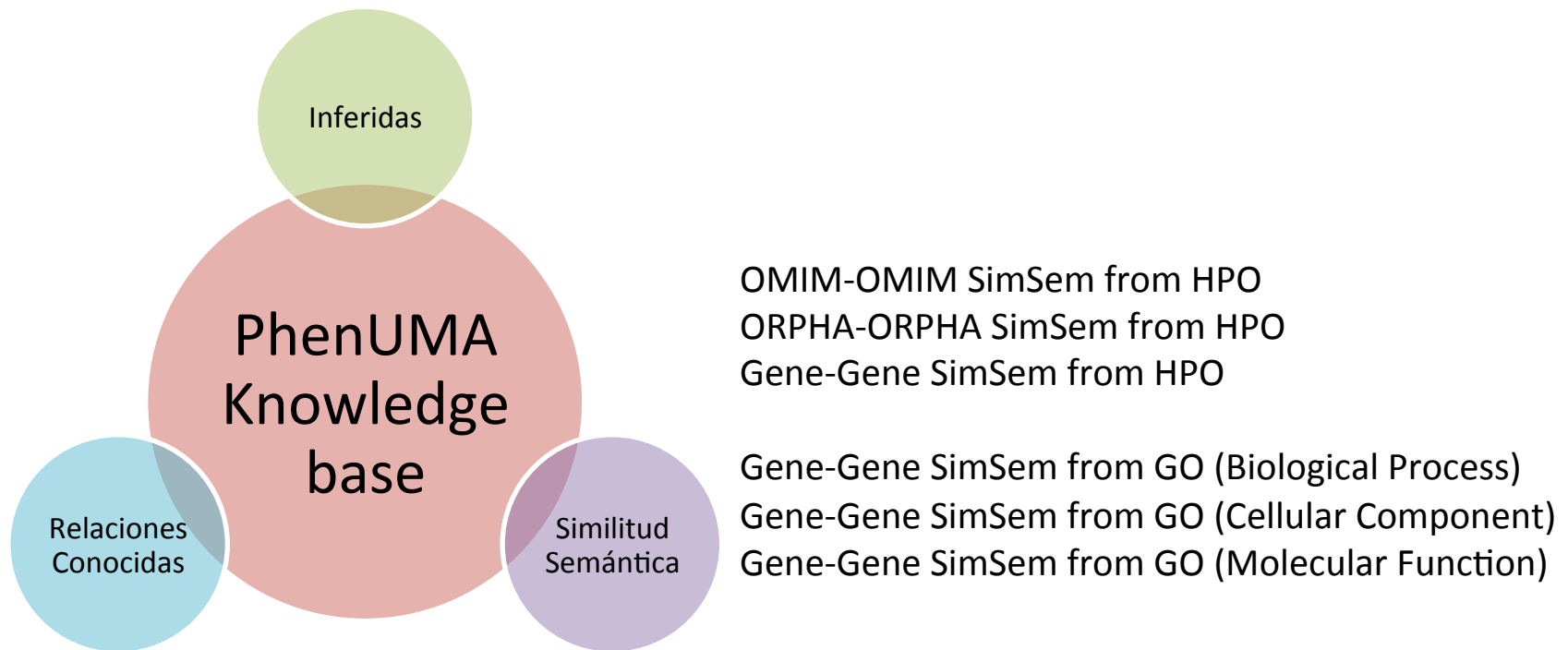
Knowledge Base : Similitud Semántica

- 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:



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	Similitud Semántica
	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		
OMIM-OMIM SimSem from HPO		

Tipos de Redes

Entrada	Tipos de Redes	
Enfermedades OMIM	Gene-OMIM Known from OMIM	Conocidas
	OMIM-OMIM Inferred from Genes	Inferidas
	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)		
OMIM-OMIM SimSem from HPO		

Tipos de Redes

Entrada	Tipos de Redes	
Enfermedades Orphanet	Gene-ORPHA Known from OMIM	Conocidas
	ORPHA-ORPHA Inferred from Genes	Inferidas
	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	Similitud Semántica

Entrada a PhenUMA → 3.Opciones de la Red de Salida → Tipos de Redes

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.

Entrada	Tipos de Redes
Fenotipos	Genes
	Enfermedades (OMIM)
	Enfermedades raras (Orphanet)

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

[Main](#) [Tutorial](#) [How to cite us](#) [Contact us](#)



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

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

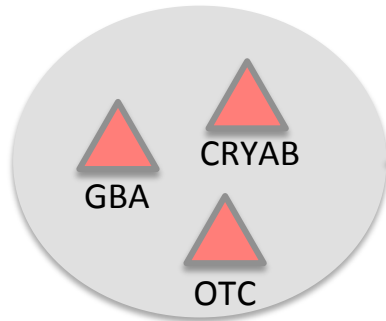
Input List:

[Load From File](#)

Construcción de la Red

Construcción de Redes

1. Genes de Interés



2. Tipo de Red de Salida:

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

3. Nivel de Confianza

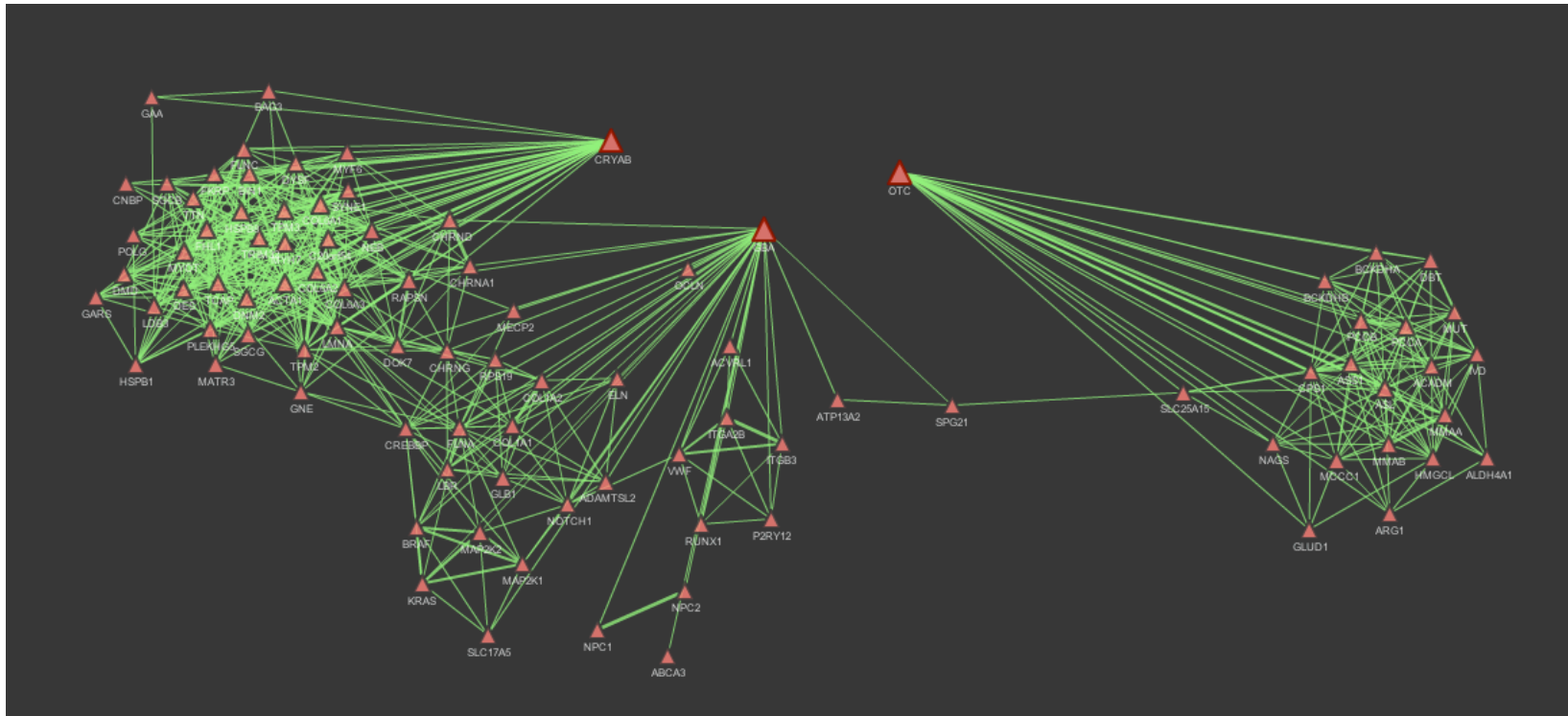
Ej: Low

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

Red Inicial
(Relaciones Fenotípicas)

Construcción de Redes

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



Knowledge Base : Construcción de Redes

