

PhenUMA Input

1 Input type

The screenshot shows the PhenUMA input interface. At the top, there are four tabs: "Genes", "OMIM (Genes/Diseases)", "Orphan Diseases", and "Phenotype". The "Genes" tab is selected. Below the tabs, there is a text input field containing "OTC". Below the input field, there are four example labels: "Example 1 Genes", "Example 2 OMIM disease", "Example 3 Orphan disease", and "Example 4 Phenotypes". Below the examples, there is a section for "Output Network:" with a dropdown menu showing "ORPHA-ORPHA Semantic Similarity from HPO" and a "Confidence:" dropdown menu showing "Low". A "Build Network" button is located at the bottom right. Annotations include a red arrow pointing to the "Genes" tab, a blue arrow pointing to the input field, and a green arrow pointing to the "Confidence:" dropdown menu.

Write a list of genes i.e. GBA, OCT or 2629, 5009:

OTC

Example 1 Genes Example 2 OMIM disease Example 3 Orphan disease Example 4 Phenotypes

Output Network: ORPHA-ORPHA Semantic Similarity from HPO Confidence: Low

Build Network

2. Input Data

3. Output network options

A network graph visualization consisting of numerous grey circular nodes connected by thin grey lines, forming a complex web-like structure. It is positioned in the top right corner of the slide, partially overlapping the black header bar.

1. Input Options

- Input type:
 - Genes
 - OMIM gene/diseases
 - Orphan Diseases
 - Phenotypes

Identifiers for Genes

- A different identifier must be used for each input type.

- **Allowed identifiers for genes:**

- Code Entrez (Recommended) → 1410,5009
- Official Symbol → CRYAB, OTC
- HGNC → HGNC:2389, HGNC:8512
- MIM → MIM:123590, MIM:300461
- Ensembl → ENSG00000109846, ENSG00000036473
- Orphanum → ORPHA:120832, ORPHA:124033



- **It can not be used:**

- Full Name: crystallin, beta A1
- Synonym: CRYB1, CTRCT10
- Protein: Beta-crystallin A3



Identifiers for Genes

A network graph visualization consisting of numerous grey circular nodes connected by thin grey lines, forming a complex web-like structure. The nodes are distributed across the top right portion of the slide, with a higher density of connections on the right side.

How to get the identifiers?

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/>

Identifiers for Genes: NCBI

The screenshot displays the NCBI website interface. At the top, there is a navigation bar with 'NCBI Resources' and 'How To' menus, and a 'Sign in to NCBI' link. The main content area features a search bar and a 'Search' button. A dropdown menu is open, listing various databases and resources, with 'Gene' highlighted. The background shows a 'Welcome to NCBI' message, a 'Twitter feed' widget, and a 'Popular Resources' section with links to PubMed, Booksshelf, PubMed Central, PubMed Health, BLAST, Nucleotide, Genome, SNP, Gene, Protein, and PubChem. Below this is an 'NCBI Announcements' section with several news items.

- ✓ 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

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: NCBI

Entrez/Gene ID

Official Symbol

Other identifiers

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 <small>provided by HGNC</small>
Official Full Name	crystallin, beta A1 <small>provided by HGNC</small>
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 eye lenses 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]

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Related information

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: Ensembl

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

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

Search: for
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

Human (GRCm37) **Mouse** (GRCm38) **Zebrafish** (Zv9)

★ [Log in to customize this list](#)

All genomes

[View full list of all Ensembl species](#)

Other species are available in [Ensembl Pre!](#) and [EnsemblGenomes](#)

ENCODE data in Ensembl

Variant Effect Predictor

New to Ensembl?

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- [Add custom tracks](#) using our 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 our 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? 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.

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: Ensembl

Results for the search: "Beta-crystallin A3"

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

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 Human...". Below the navigation bar, the species is set to "Human (GRCh37)". 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 "Results Summary" and states: "Your search of Human with 'Beta-crystallin A3' returned the following results:". Below this, there are two summary tables:

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

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

At the bottom of the page, there is a footer with the text "Ensembl release 71 - April 2013 © WTSI / EBI" and links for "About Ensembl", "Privacy Policy", and "Contact Us". A "Permanent link - View in archive site" link is also present.

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: Ensembl

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 is located on the right. Below the navigation bar, there is a sidebar on the left with options like 'Search Ensembl', 'Configure this page', 'Add your data', 'Export data', 'Bookmark this page', and 'Share this page'. The main content area displays the 'Result in Detail' for a query: '1 Gene matches your query ('Beta-crystallin A3') in Human'. The gene identified is 'CRYBA1'. The details include: Description: 'crystallin, beta A1 [Source:HGNC Symbol;Acc:2394] [Type: protein coding Ensembl/Havana merge]'; Gene ID: 'ENSG00000108255' (highlighted with an orange box); 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'. An orange arrow points from the highlighted Ensembl ID to a box labeled 'Ensembl ID' below the screenshot.

Ensembl ID

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for 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, with 'MT' for mitochondrial DNA. 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'. The search options include 'Search symbols, keywords or IDs for:' with radio buttons for 'equal', 'begin', and 'contain' (selected). The 'Display' dropdown is set to '50 hits'.

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: HGNC

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

The screenshot shows the HGNC (HUGO Gene Nomenclature Committee) website. The header includes the HGNC logo and a search bar. A navigation menu contains links for Home, Search Genes, Downloads, Gene Families, HCOP, Useful Links, About, and Contact Us, along with a Request Symbol button. The main content area is titled 'Quick Gene Search' and features a search form with the following options: 'Search symbols, keywords or IDs for:', 'Results that' (radio buttons for equal, begin, and selected 'contain'), and 'Display' (a dropdown menu set to 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', and 'Next Page'. The search results are displayed in a table with 3441 total hits. The table has four columns: 'Approved Symbol', 'Approved Name', 'Location', and 'Best Match'. The first row shows the symbol 'CRYBA1' for 'crystallin, beta A1' located at '17q11.2-q12' with a best match of 'Approved Name: crystallin, beta A1'. Other rows include 'CRYBG3', 'CRYBA2', 'CRYBA4', 'CRYBB1', 'CRYBB2', and 'CRYBB2P1'.

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: HGNC

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

The screenshot shows the HGNC Gene Symbol Report for the gene CRYBA1. The report is organized into several sections: Symbol, Homologs, Nucleotide Sequences, Gene Resources, Protein Resources, and Clinical Resources. Callouts from the left point to specific fields: 'Symbol' points to the 'Approved Symbol' field (CRYBA1), 'HGNC ID' points to the 'HGNC ID' field (HGNC:2394), and 'Others' points to the 'Gene Resources' section, which lists various databases and their corresponding identifiers for the gene.

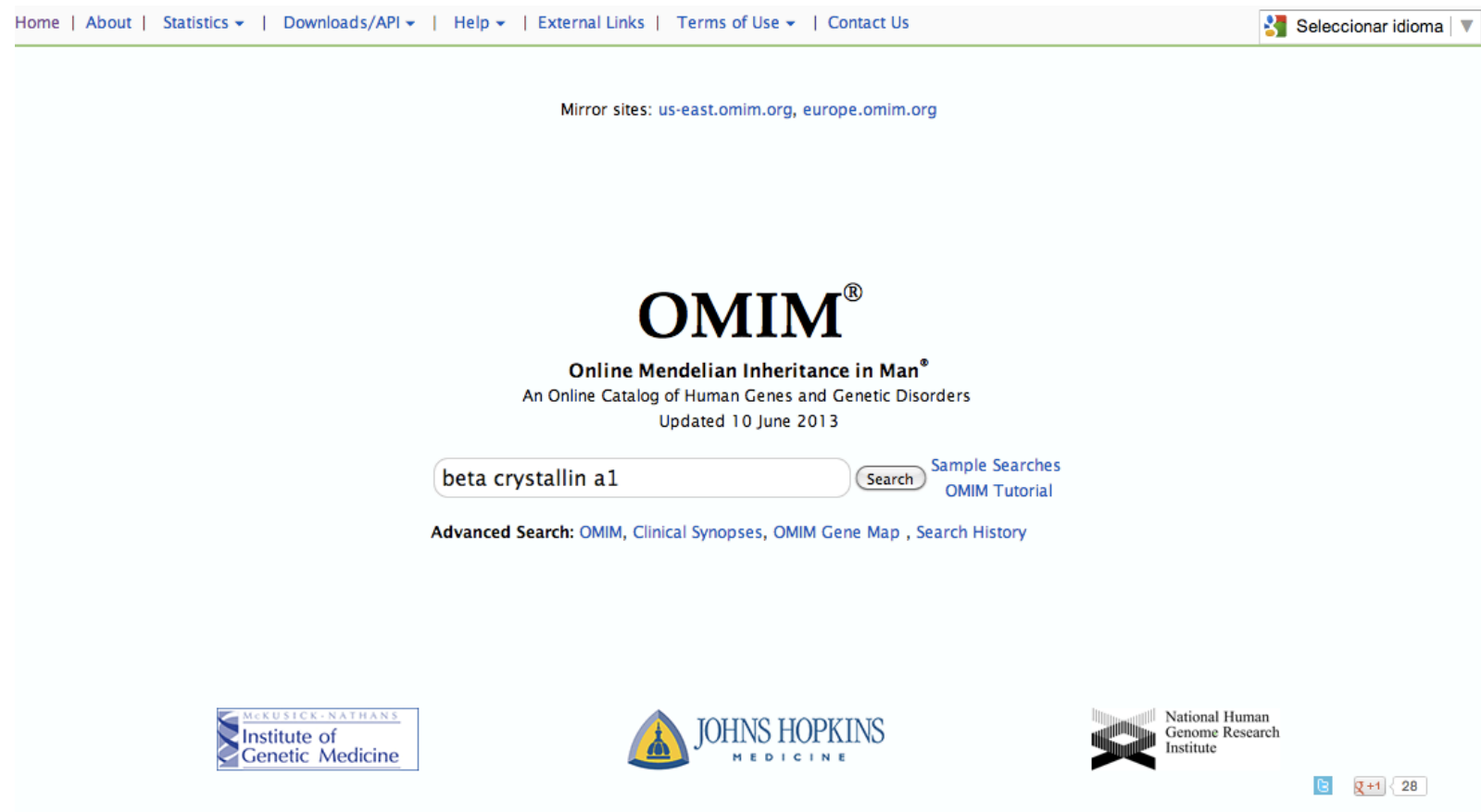
Field	Value
Approved Symbol	CRYBA1
Approved Name	crvstallin, beta A1
HGNC ID	HGNC:2394
Previous Symbols & Names	CRYB1
Synonyms	"eye lens structural protein"
Locus Type	gene with protein product
Chromosomal Location	17q11.2-q12

Section	Resources
NUCLEOTIDE SEQUENCES	RefSeq: NM_005208
	CCDS: CCDS11249.1
	Vega: OTTHUMG00000132729
	Entrez Gene: 1411
GENE RESOURCES	Ensembl: ENSG00000108255
	UCSC: uc002hdw.3
	Vega: OTTHUMG00000132729
	NCBI Sequence Viewer
	Ensembl Genome Browser
PROTEIN RESOURCES	UniProtKB: P05813
	InterPro
	OMIM
	GeneTests
CLINICAL RESOURCES	Orphanet
	DECIPHER
	COSMIC
	LSDB: LOVD - Leiden Open Variation Database
	Genetic Testing Registry
	Mouse Symbol: Cryba1
Rat Symbol: Cryba1	

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for 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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PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: MIM

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

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

Sort by: Relevance Date updated

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

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	

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: MIM

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

The screenshot shows the OMIM website interface. At the top, there is a navigation bar with links: Home, About, Statistics, Downloads/API, Help, External Links, Terms of Use, and Contact Us. A language selector 'Seleccionar idioma' is on the right. The search bar contains 'beta crystallin a1' and has a 'Search' button. To the right of the search bar, there are options for 'Sort by: Relevance' (selected) and 'Date updated'. Below the search bar, there is a link for 'Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map' and a 'Toggle: search terms highlighted, changes highlighted' option. The search results show the OMIM ID '*123610' and the gene name 'CRYSTALLIN, BETA-A1; CRYBA1'. Underneath, there are 'Alternative titles; symbols' listed: 'CRYSTALLIN, BETA-1; CRYB1' and 'CRYSTALLIN, BETA-A1 / A3'. A callout box highlights the 'HGNC Approved Gene Symbol: CRYBA1'. Below this, the 'Cytogenetic location' is '17q11.2' and the 'Genomic coordinates (GRCh37)' are '17:27,573,874 - 27,581,511 (from NCBI)'. At the bottom, there is a section for 'Gene Phenotype Relationships' with a table:

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

On the right side of the page, there is a 'Table of Contents - *123610' section with a list of external links: Genome, DNA, Protein, Gene Info, Clinical Resources, Variation, Animal Models, and Cellular Pathways.

OMIM ID → *123610

Symbol → HGNC Approved Gene Symbol: CRYBA1

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Genes: Orphanum

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

The screenshot shows the Orphanet website homepage. At the top, there is a navigation bar with language options: FR, EN, ES, DE, IT, PT. The main header features the Orphanet logo, the tagline "The portal for rare diseases and orphan drugs", and the slogan "Rare diseases are rare, but rare disease patients are numerous". Below the header is a banner with several images related to medicine and research. A central navigation bar contains the text "Access our Services" and a search box with an "OK" button. Below this, there are two columns of service links. The left column includes: "Inventory, classification and encyclopaedia of rare diseases, with genes involved", "Assistance-to-diagnosis tool", "Emergency guidelines", "Inventory of orphan drugs", "Directory of medical laboratories providing diagnostic tests", and "Directory of expert centres". The right column includes: "Directory of ongoing research projects, clinical trials, registries and biobanks", "Directory of patient organisations", "Directory of professionals and institutions", "Newsletter", and "Collection of thematic reports: Orphanet Reports Series". Below the service links are three green buttons: "Read Orphanet reports", "Contribute to Orphanet", and "Download Orphanet data". The "Read Orphanet reports" button is highlighted with an orange box and an arrow pointing to a larger orange box at the bottom of the slide containing the text "Finding genes or diseases". The "Contribute to Orphanet" button contains links for "Register your activity" and "Sponsor Orphanet". The "Download Orphanet data" button contains a link for "Orphadata". On the right side of the page, there are sections for "Newsletter", "Other documents", "Other rare diseases websites", and "Events". The "Events" section shows an event for "JULY 1 Myasthenia 2013" from "1 - 2 July 2013, Paris, France".

Finding genes or diseases

PhenUMA Input → 1. Input Options → Identifiers for 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 navigation links (Homepage, Help, Contact us). The main navigation bar includes 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 this, 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 entry for 'CRYBA1 - Crystallin, beta A1' with various identifiers listed.

Type of query: Gene

Finding form: type of identifier

Output: Orphanum, OMIM, HGNC, Ensembl....

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

Identifiers for Diseases: OMIM

- OMIM
 - <http://www.omim.org/>
 - Search by disease name:
 - 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

PhenUMA Input → 1. Input Options → Identifiers for Diseases

Identifiers for Diseases: Orphanum

- Rare diseases

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 is set to "EN". A navigation menu includes categories like "Rare diseases", "Orphan drugs", "Expert centres", etc. The main content area shows a search for "Friedreich ataxia" with search options like "Disease name", "Gene name or symbol", etc. Below the search, there is a table of identifiers for Friedreich ataxia.

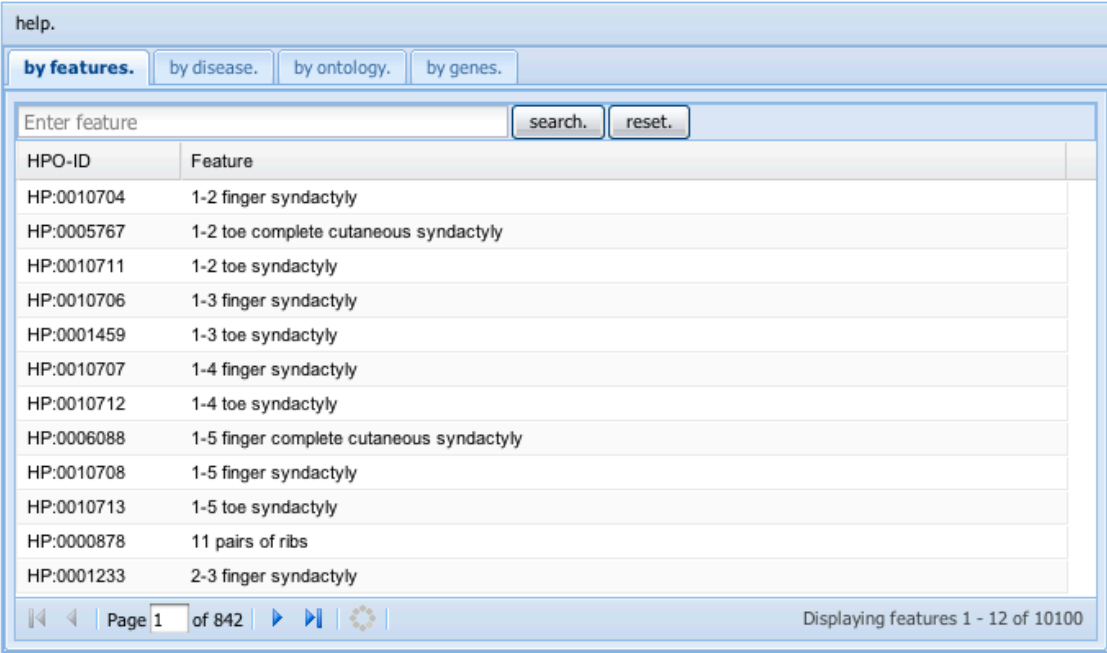
Orpha number	: ORPHA95	ICD-10	: G11.1
Synonym(s)	: -	OMIM	: 229300 601992
Prevalence	: 1-9 / 100 000	UMLS	: C0016719
Inheritance	: Autosomal recessive	MeSH	: D005621
Age of onset	: Childhood	MedDRA	: 10017374
		SNOMED CT	: 10394003

PhenUMA Input → 1. Input Options → Identifiers for Diseases

Phenotypes

- 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

PhenUMA Input → 1. Input Options → Identifiers for Diseases