

# PhenUMA Input

1 Input type

Genes

OMIM (Genes/Diseases)

Orphan Diseases

Phenotype

*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

# 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 (Recomended) → 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

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/>

Orphanet

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

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

# Identifiers for Genes: NCBI

NCBI Resources How To Sign in to NCBI

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

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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 inside look at what's new at NCBI.

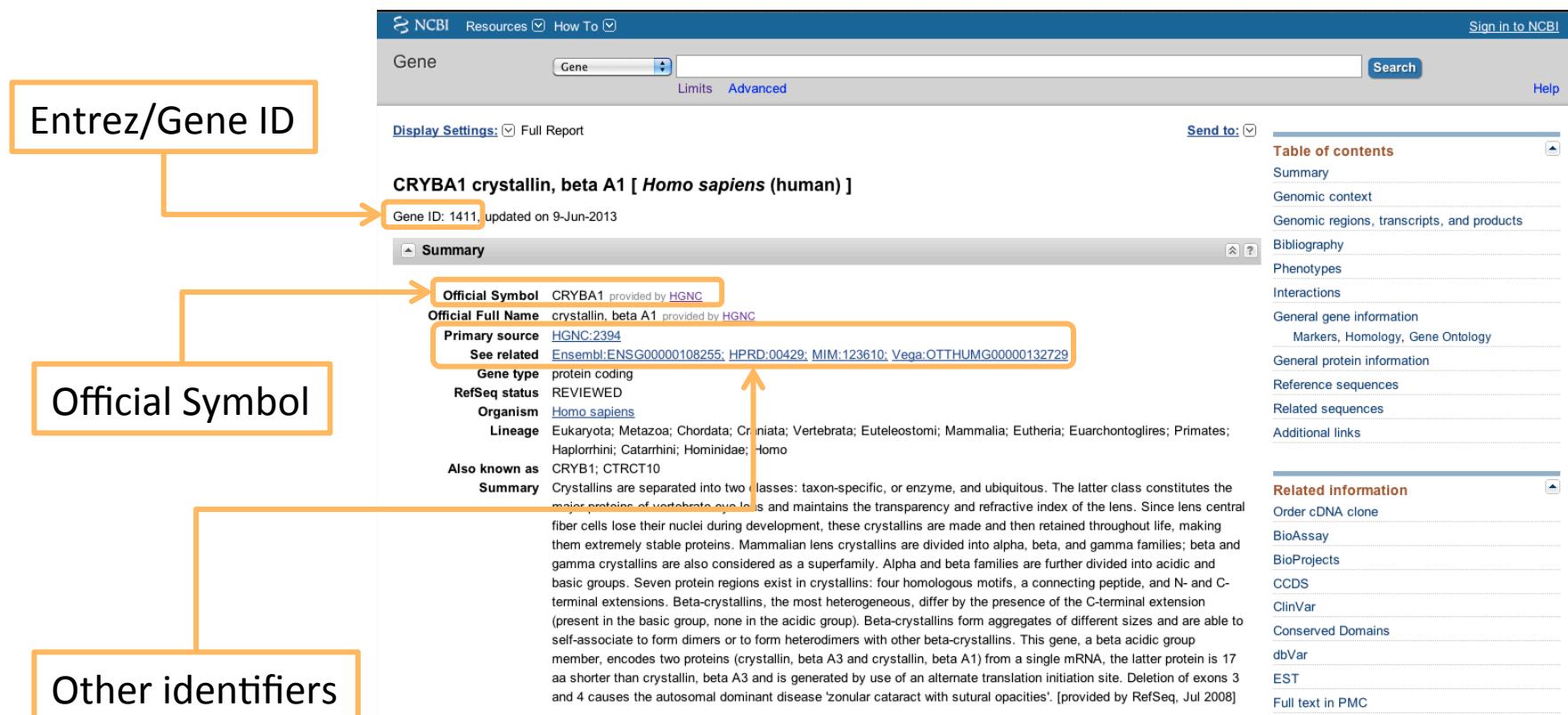
Come to the NCBI Discovery Workshops on February 4&5! 16 Jan 2013 Spaces are still available for the free, 2-day Discovery Workshops to be held on Feb 4-5.

New version of Genome Workbench available 06 Sep 2012 An integrated, downloadable application for viewing and analyzing genomic data

More...

PhenUMA Input → 1. Input Options → Identifiers for Genes

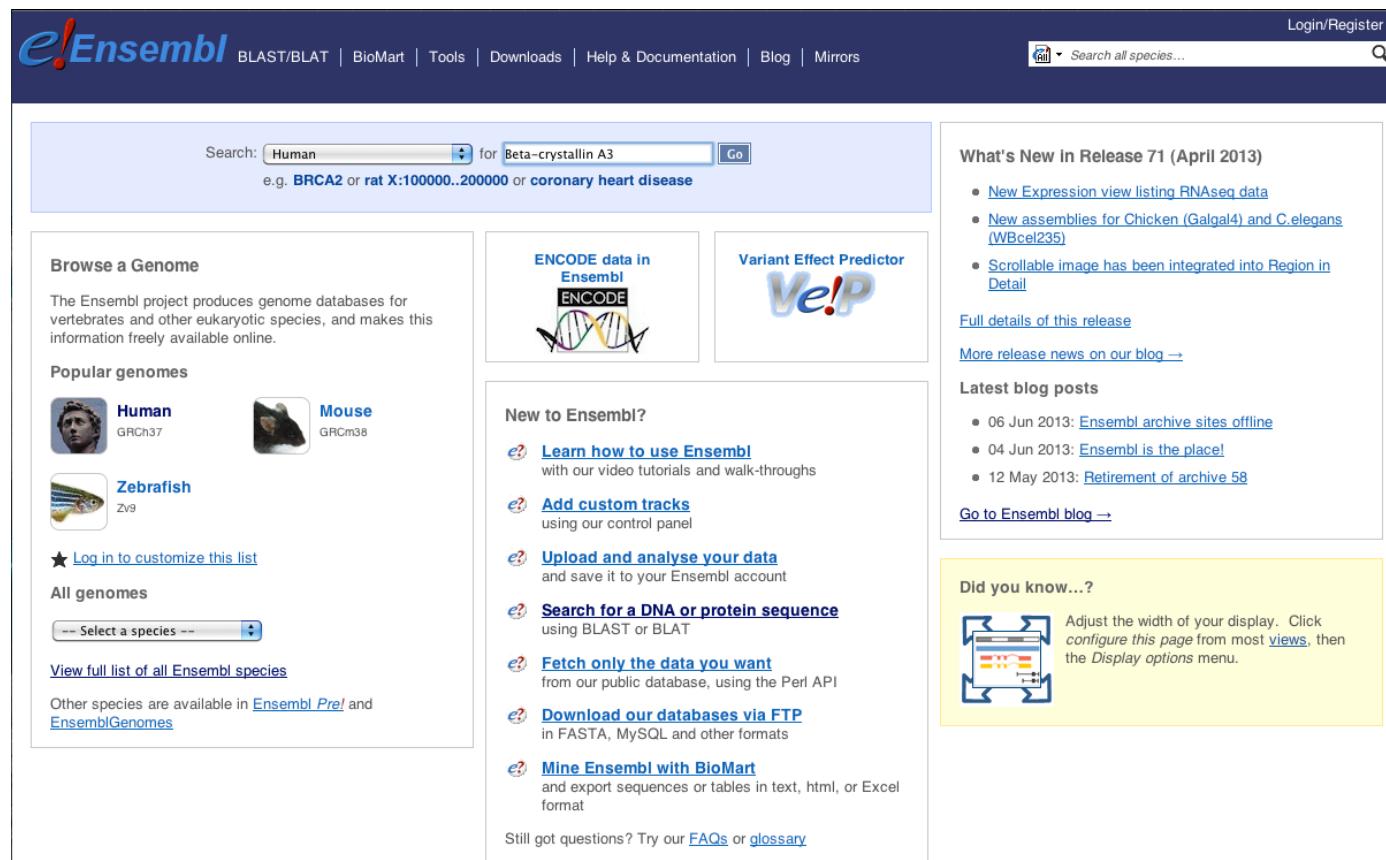
# Identifiers for Genes: NCBI



PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: Ensembl

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



The Ensembl homepage features a search bar at the top with the text "Search: Human" and "for Beta-crystallin A3". Below the search bar are sections for "Browse a Genome", "Popular genomes" (Human, Mouse, Zebrafish), and "New to Ensembl?" with links to video tutorials, custom tracks, data upload, DNA/protein search, data fetching, database download, and BioMart mining. The right side of the page includes a "What's New in Release 71 (April 2013)" section, a "Latest blog posts" section, and a "Did you know...?" section with a tip about adjusting display width.

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

The screenshot shows the Ensembl search results for the query "Beta-crystallin A3". 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 Human (1)	1
Transcript Human (1)	1

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

Ensembl release 71 - April 2013 © WTSI / EBI

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for 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 for 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 "1 Gene matches your query ('Beta-crystallin A3') in Human". The gene name "CRYBA1" is displayed, along with its description as "crystallin, beta A1 [Source:HGNC Symbol;Acc:2394] [Type: protein coding Ensembl/Havana merge]". The gene ID "ENSG00000108255" is highlighted with an orange box and an orange arrow points to it from the label "Ensembl ID" at the bottom left. Below the ID are the location "17:27573481-27581512:1" and a link to the "Variation Table". The page footer includes links to Ensembl release 71 (April 2013), WTSI / EBI, About Ensembl, Privacy Policy, and Contact Us.

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: 17:27573481-27581512:1

Variations: Variation Table

Source: e71

Ensembl release 71 - April 2013 © WTSI / EBI

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Permanent link - View in archive site

Ensembl ID

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. At the top, there is a navigation bar with links for Home, Search Genes, Downloads, Gene Families, HGCP, Useful Links, About, Contact Us, and Request Symbol. A search bar labeled "Search Genes" is located at the top right. Below the navigation bar, there is a diagram of human chromosomes numbered 1 through 22, X, and Y. To the right of the chromosomes, a text block provides information about the HGNC's mission to assign unique gene symbols and names to over 37,000 human loci, with around 19,000 being protein coding. It also mentions [genenames.org](http://genenames.org) as a curated online repository. A "Quick Gene Search" box is visible on the right, containing a search input field with the text "crystallin|beta A1" and a search button.

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: HGNC

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



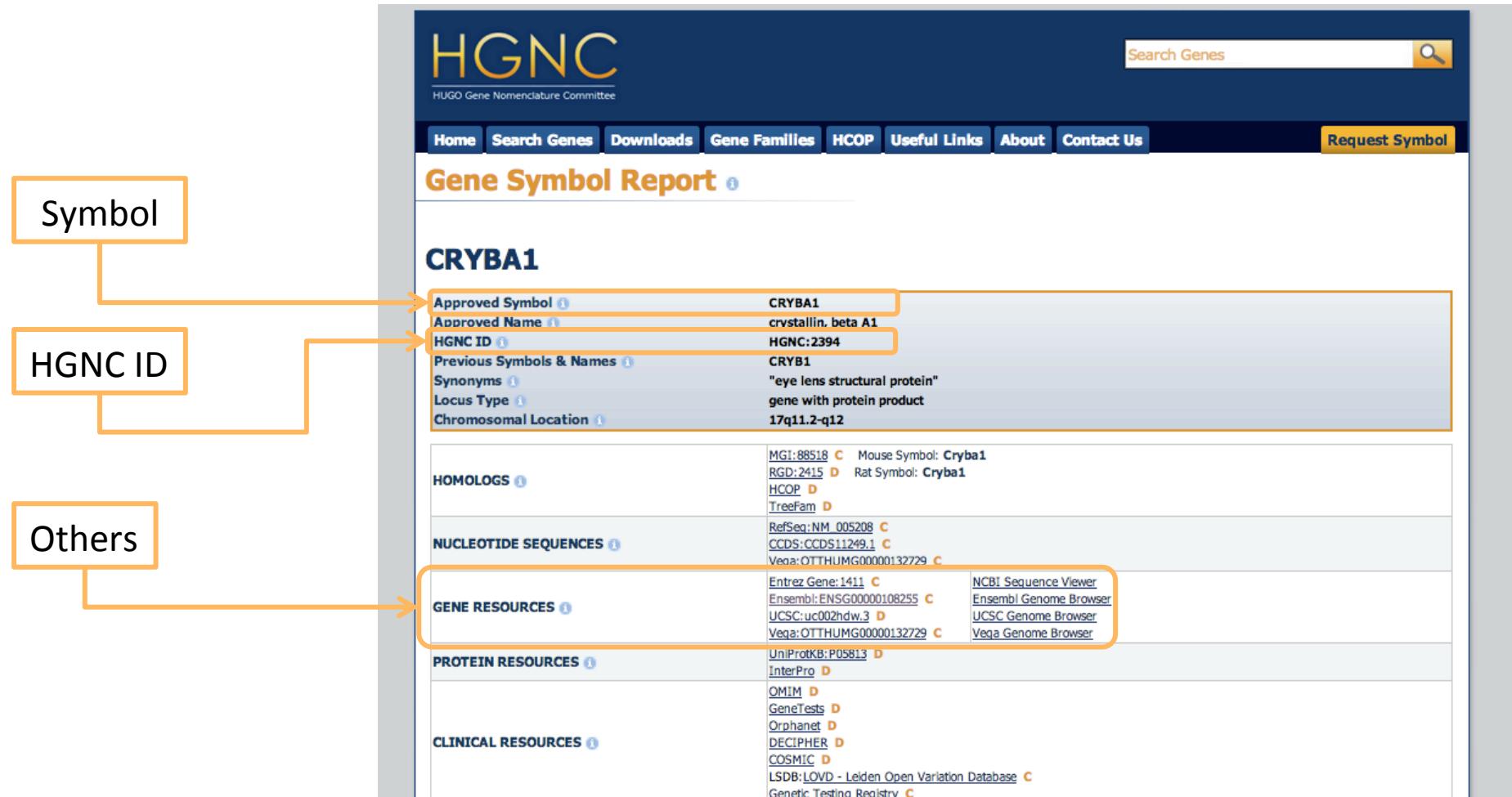
The HGNC website interface. At the top, there's a dark blue header bar with the HGNC logo and a search bar labeled "Search Genes". Below the header is a navigation menu with links to Home, Search Genes, Downloads, Gene Families, HGCP, Useful Links, About, Contact Us, and Request Symbol. A "Quick Gene Search" section is prominently displayed, featuring a search input field containing "crystallin beta A1" and a search button. Below this, a table lists gene entries, with the first few rows shown:

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: HGNC

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



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](http://us-east.omim.org), [europe.omim.org](http://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](#)



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PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: MIM

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

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

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: MIM

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

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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

**OMIM ID**

\*123610

**Symbol**

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
<a href="#">17q11.2</a>	Cataract 10, multiple types	600881

Table of Contents - \*123610  
External Links:  
▶ Genome  
▶ DNA  
▶ Protein  
▶ Gene Info  
▶ Clinical Resources  
▶ Variation  
▶ Animal Models  
▶ Cellular Pathways

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identifiers for Genes: Orphanum

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

Languages: FR EN ES | DE | IT | PT

Rare diseases are rare, but rare disease patients are numerous

Homepage About Orphanet Help Contact us

Access our Services

- 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
- Directory of expert centres
- Directory of ongoing research projects, clinical trials, registries and biobanks
- Directory of patient organisations
- Directory of professionals and institutions
- Newsletter
- Collection of thematic reports: Orphanet Reports Series

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

Finding genes or diseases

PhenUMA Input → 1. Input Options → Identifiers for Genes

# Identificadores de Genes: Orphanum

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

The screenshot shows the Orphanet homepage with various navigation links and a search interface. A search term 'beta crystallin a1' has been entered into the 'Gene name or symbol' field of a 'SIMPLE SEARCH' form. The 'OK' button is highlighted with an orange arrow. The search results for 'CRYBA1 - Crystallin, beta A1' are displayed, showing various identifiers and their corresponding values.

**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

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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 homepage with a navigation bar at the top. The main content area features a search form for "Friedreich ataxia". The search results table includes columns for Orpha number, Synonym(s), Prevalence, Inheritance, Age of onset, ICD-10, OMIM, UMLS, MeSH, MedDRA, and SNOMED CT.

**Orphanet**  
The portal for rare diseases and orphan drugs  
Inserm    European Union

Rare diseases are rare, but rare disease patients are numerous

Languages: FR EN ES DE IT PT  
Homepage Help Contact us

Rare diseases   Orphan drugs   Expert centres   Diagnostic tests   Research and trials   Patient organisations   Professionals and institutions   Other information

Search   Search by sign   Classifications   Genes   Encyclopaedia for patients   Encyclopaedia for professionals   Emergency guidelines

Homepage » Rare diseases » Search   Seleccionar idioma   Print

Friedreich ataxia   (\*) mandatory field   OK   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

PhenUMA Input → 1. Input Options → Identifiers for Diseases

# Phenotypes

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

The screenshot shows a web-based search interface for HPO features. At the top, there is a navigation bar with tabs: 'by features.' (which is selected and highlighted in blue), 'by disease.', 'by ontology.', and 'by genes.'. Below the navigation bar is a search input field labeled 'Enter feature' with a placeholder 'HPO-ID' and a 'Feature' dropdown. To the right of the input field are two buttons: 'search.' and 'reset.'. The main content area is a table with two columns: 'HPO-ID' and 'Feature'. The table lists 12 features from page 1 of 842, with a total of 10100 features displayed. The listed features include various types of syndactylies (e.g., 1-2 finger syndactyly, 1-2 toe syndactyly, 1-3 finger syndactyly, 1-4 finger syndactyly, 1-4 toe syndactyly) and other conditions like 11 pairs of ribs. At the bottom of the table, there are navigation icons for first, previous, next, last, and a search icon, followed by the text 'Page 1 of 842'. To the right of the table, it says 'Displaying features 1 - 12 of 10100'.

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

PhenUMA Input → 1. Input Options → Identifiers for Diseases