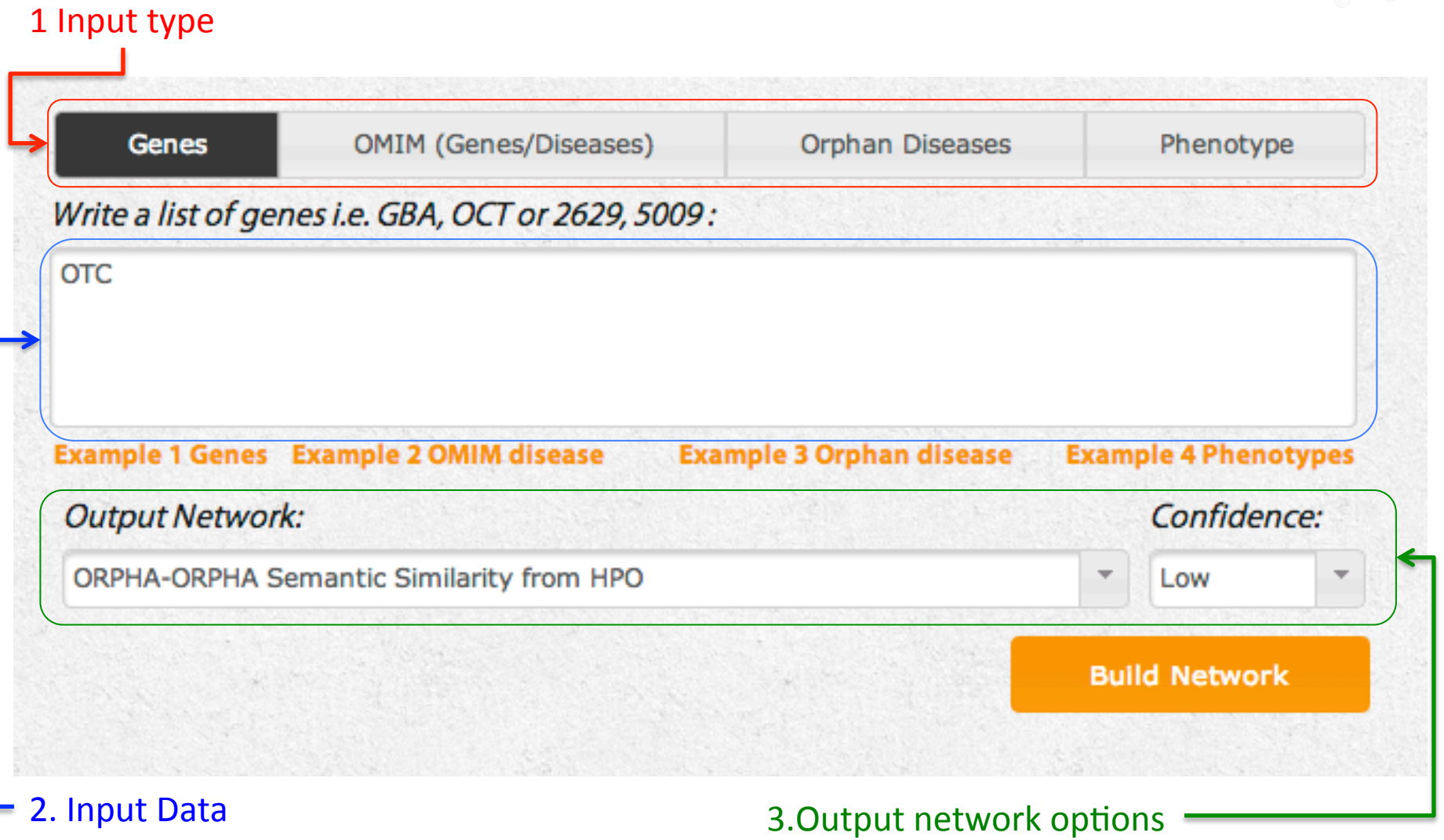
PhenUMA Input



PhenUMA Input

1. Input Options

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

Identifiers for Genes

- A different indentifier must to be used for each input type.
- **Allowed indentifiers 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

PhenUMA Input→ 1. Input Options → Identifiers for Genes

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/

Orphanum
http://www.orpha.net/

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: NCBI

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: NCBI

Entrez/Gene ID

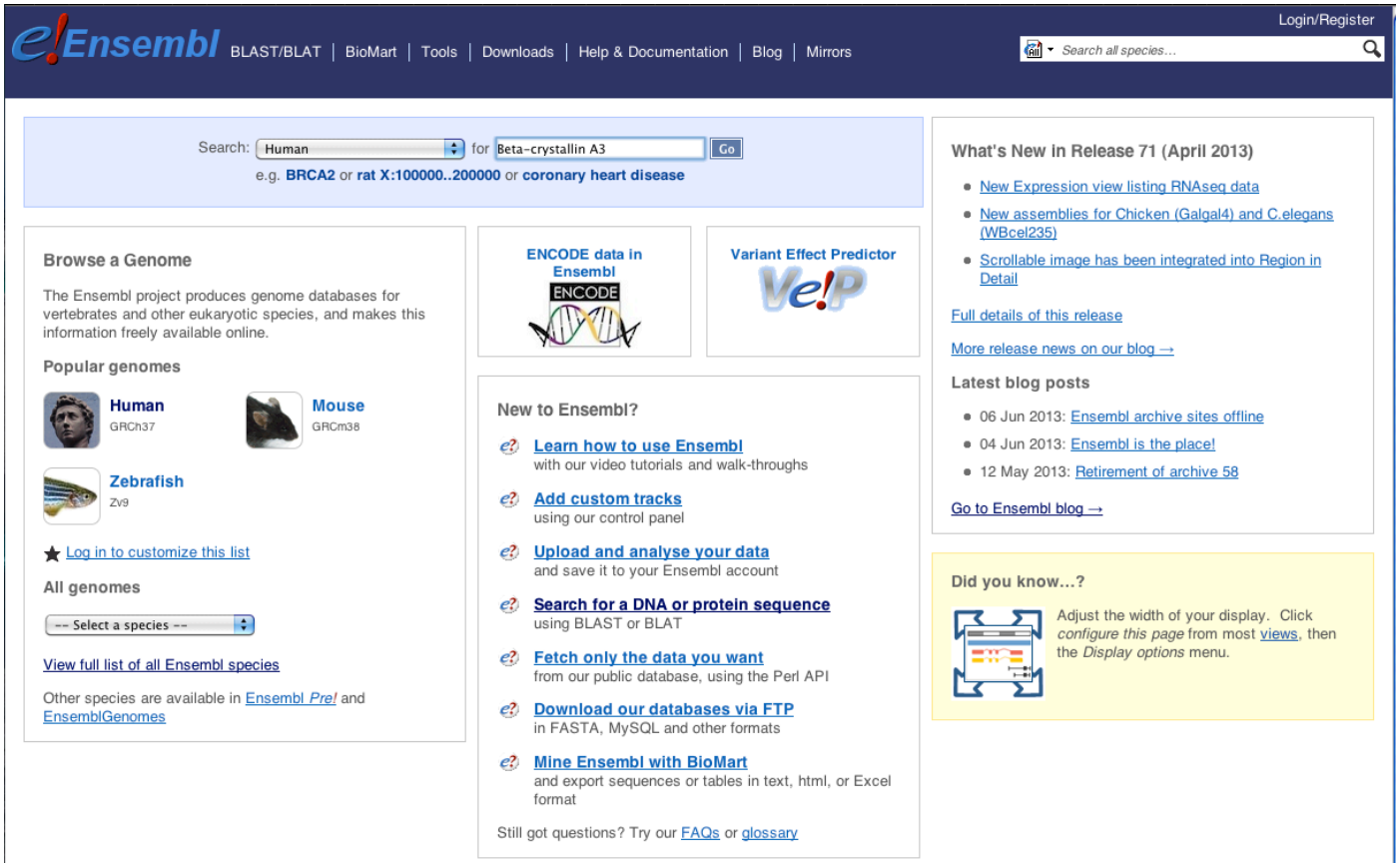
Official Symbol

Other identifiers

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: Ensembl

http://www.ensembl.org/



PhenUMA Input→ 1. Input Options → Identifiers for Genes

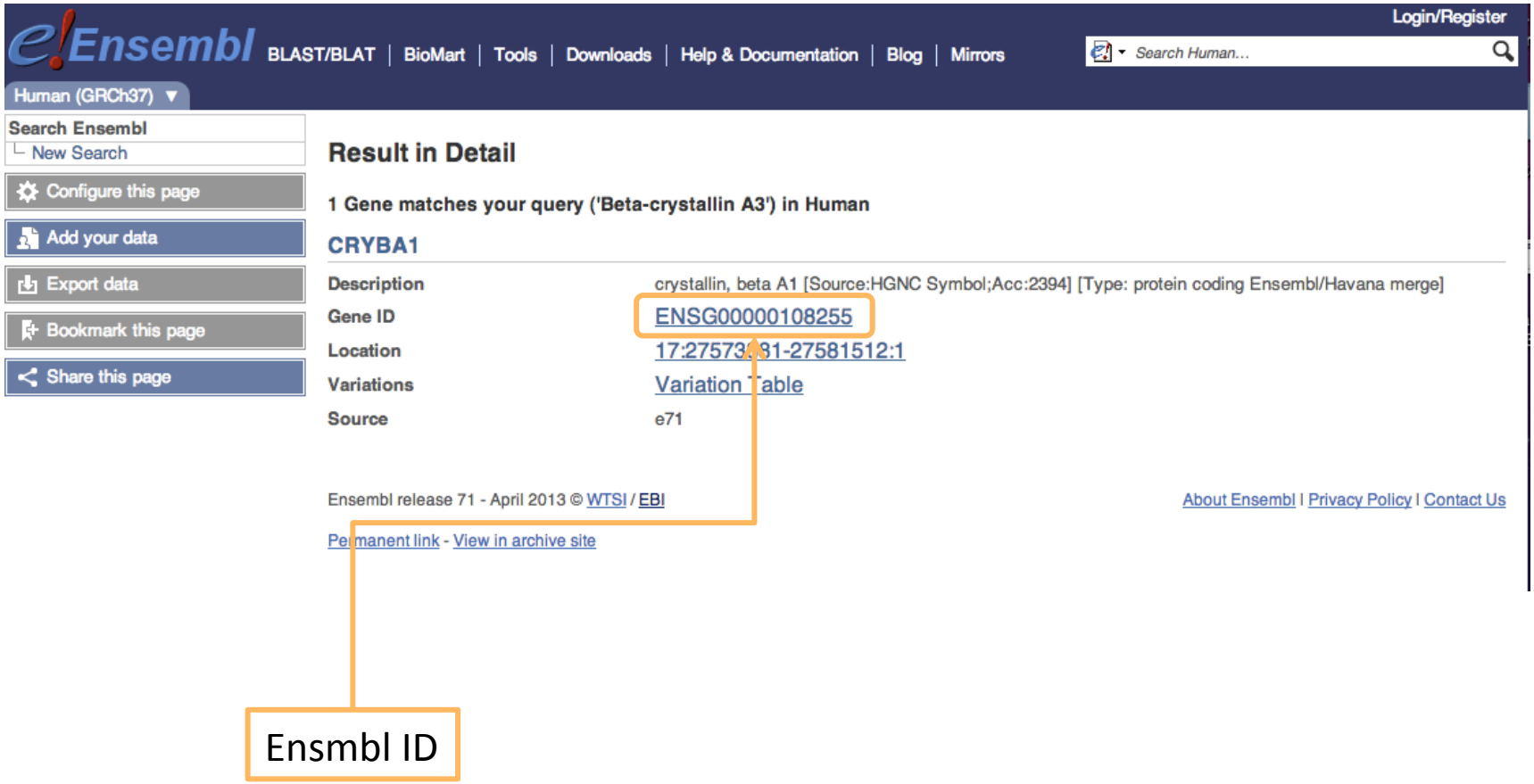
Identifiers for Genes: Ensembl

Results for the search: “Beta-crystallin A3”

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

PhenUMA Input→ 1. Input Options → Identifiers for Genes

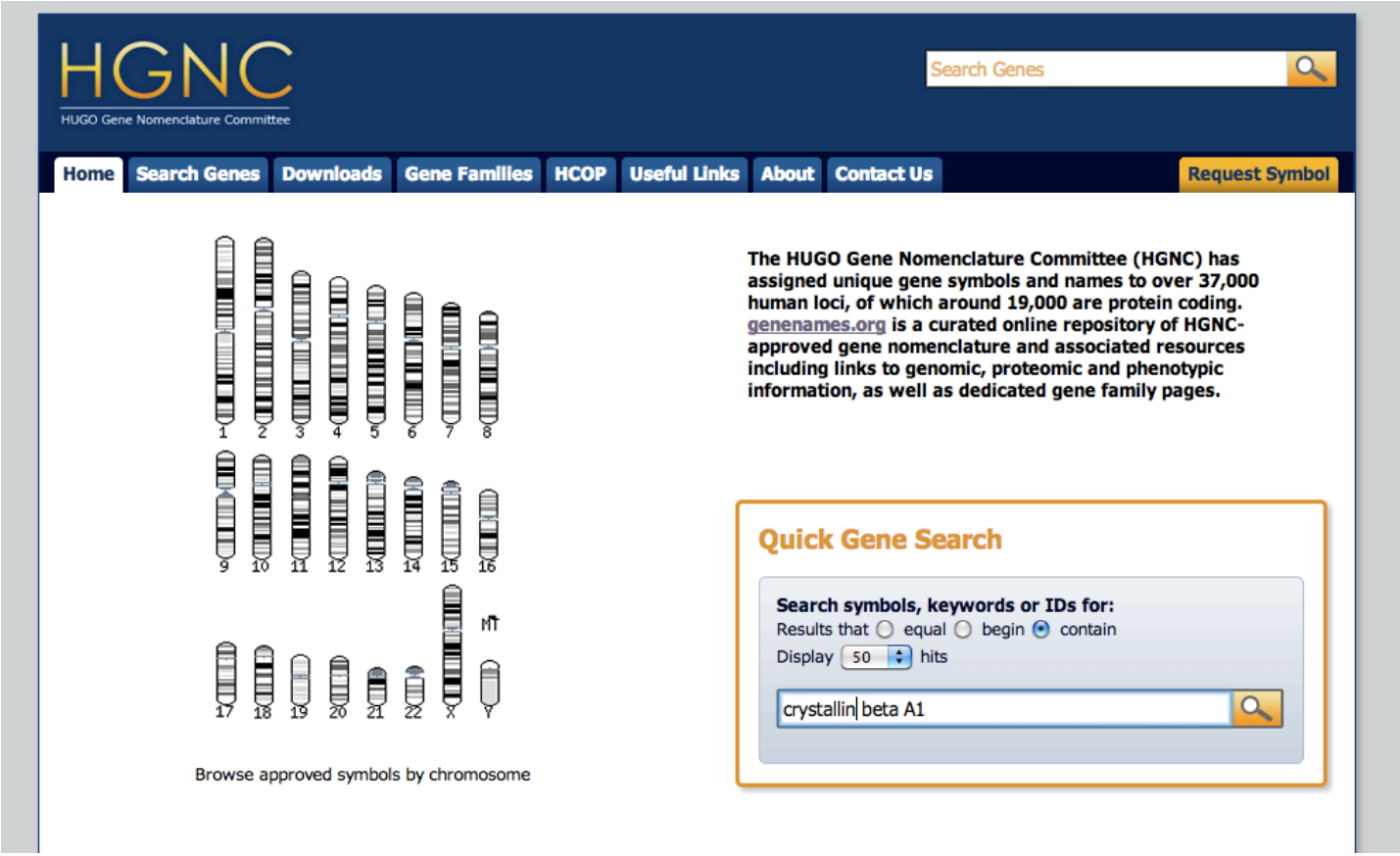
Identifiers for Genes: Ensembl



PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: HGNC

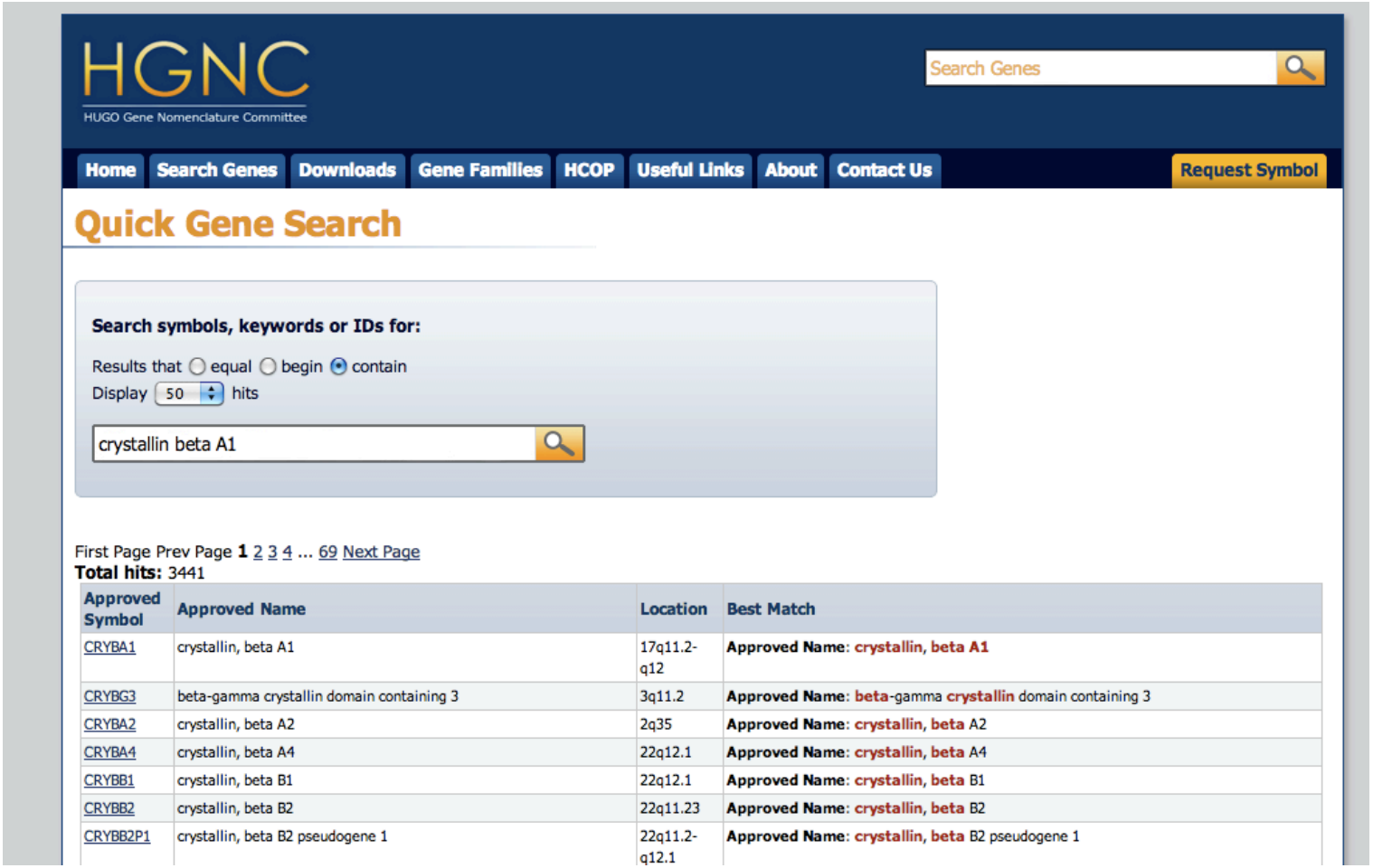
http://www.genenames.org/



PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: HGNC

http://www.genenames.org/



PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: HGNC

http://www.genenames.org/

Symbol

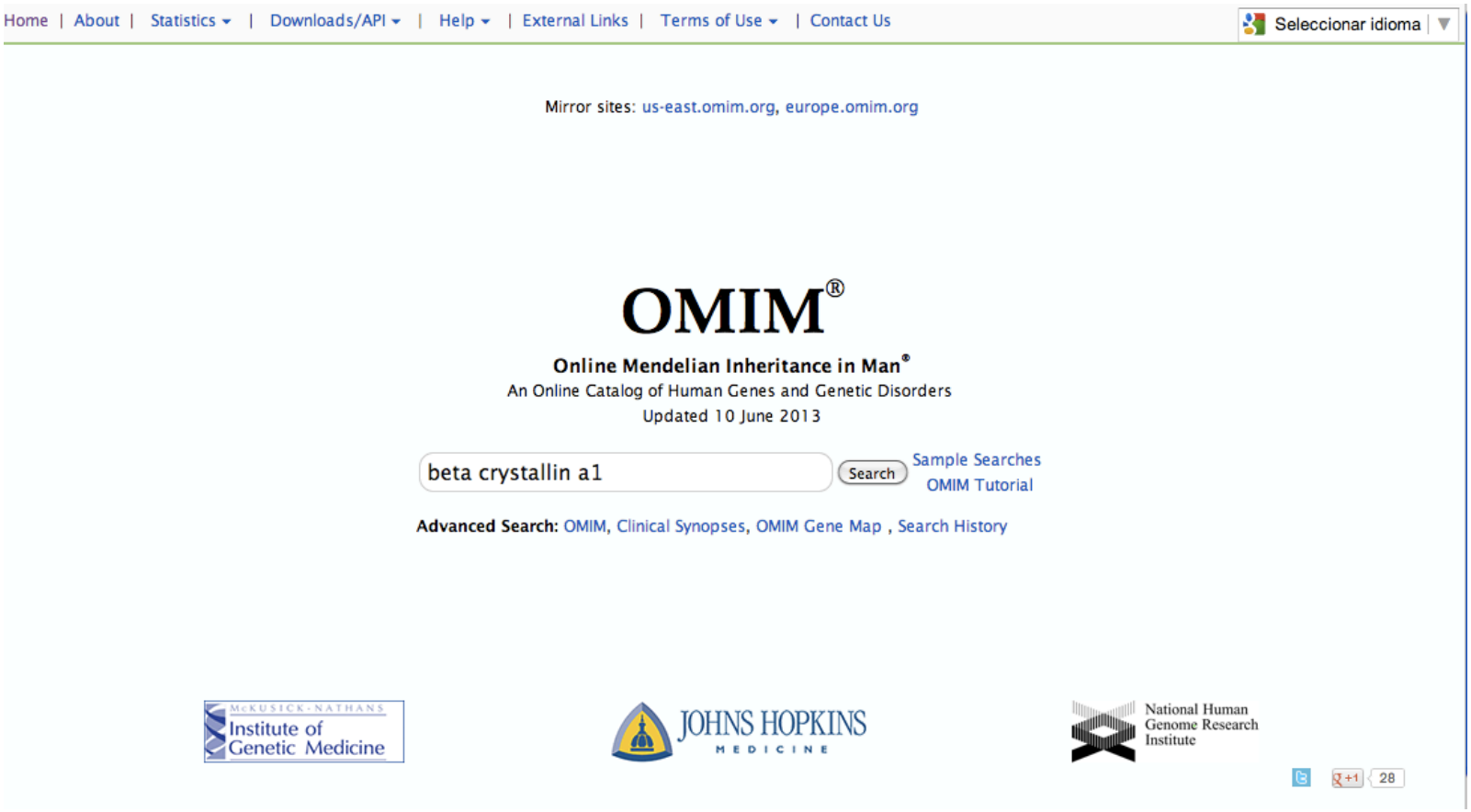
HGNC ID

Others

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: MIM

http://www.omim.org/



PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: MIM

http://www.omim.org/

PhenUMA Input→ 1. Input Options → Identifiers for Genes

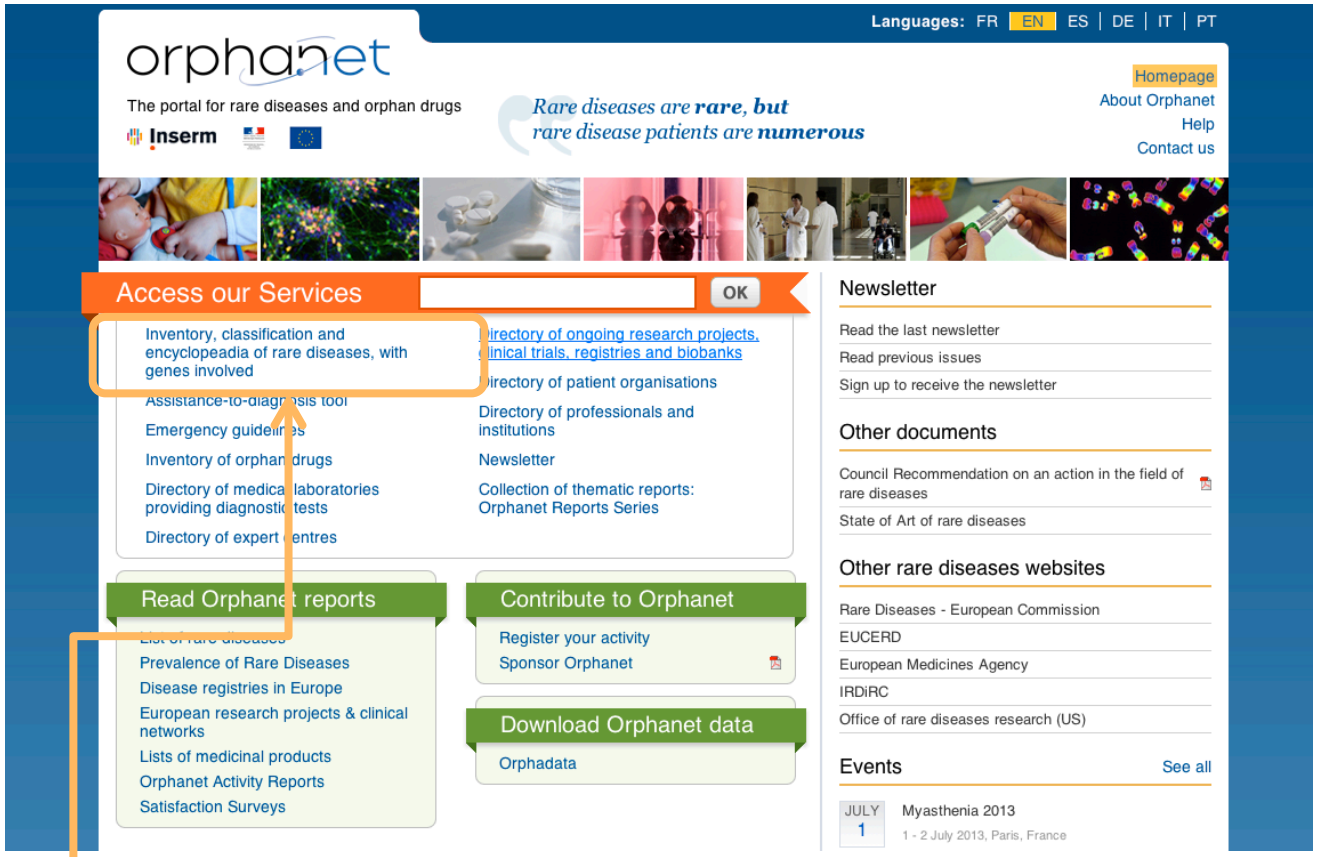
Identifiers for Genes: MIM

http://www.omim.org/

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Genes: Orphanum

http://www.orpha.net/

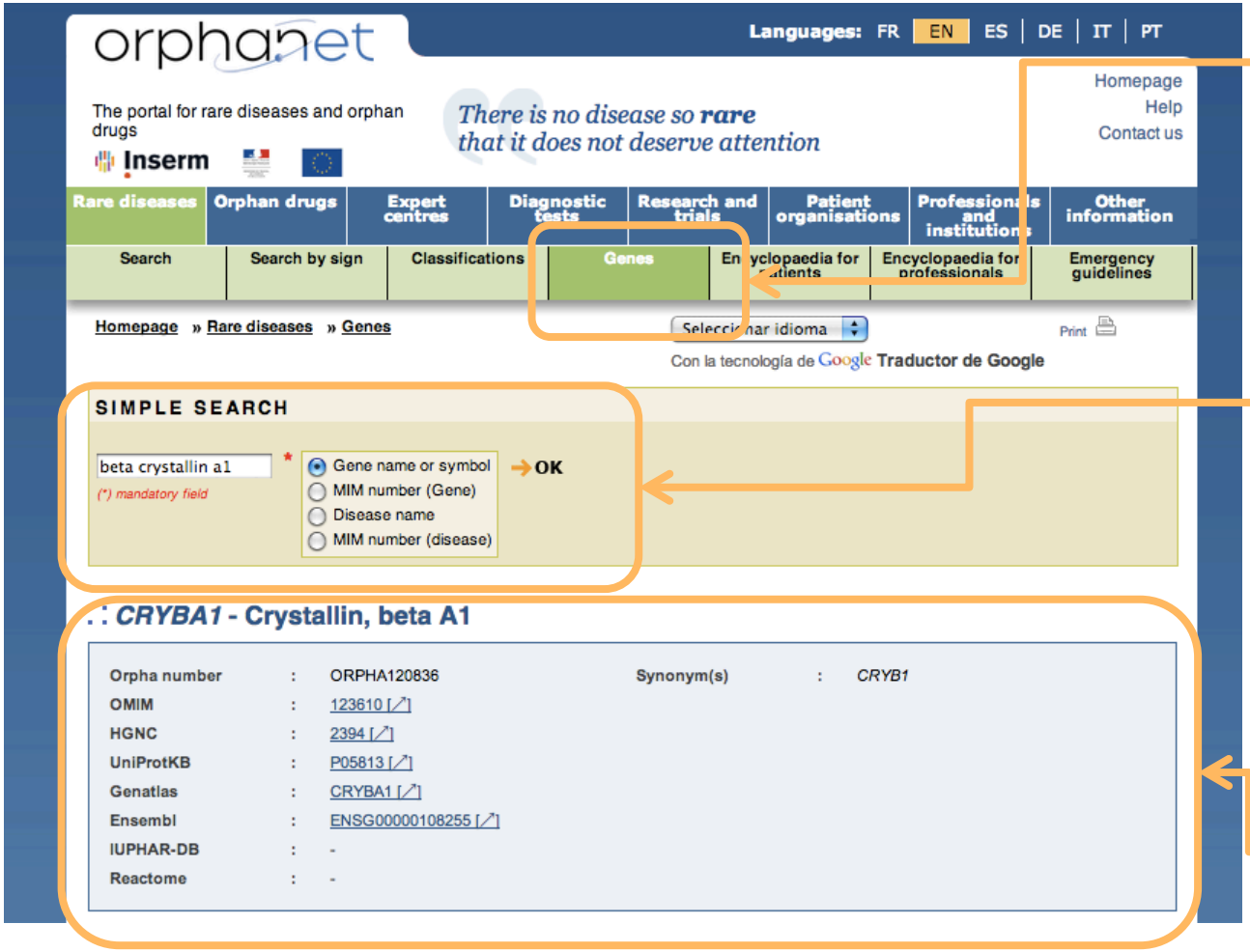


Finding genes or diseases

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Indetificadores de Genes: Orphanum

http://www.orpha.net/



Type of query: Gene

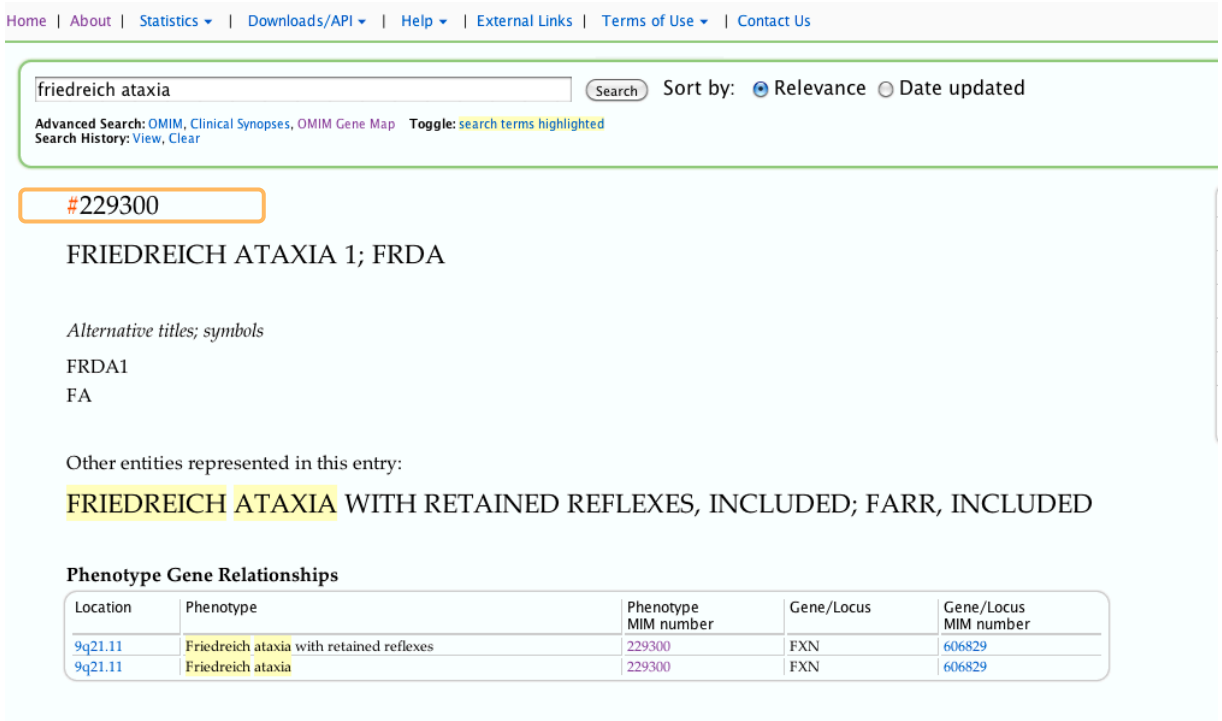
Finding form: type of identifier

Output: Orphanum, OMIM, HGNC, Ensembl….

PhenUMA Input→ 1. Input Options → Identifiers for Genes

Identifiers for Diseases: OMIM

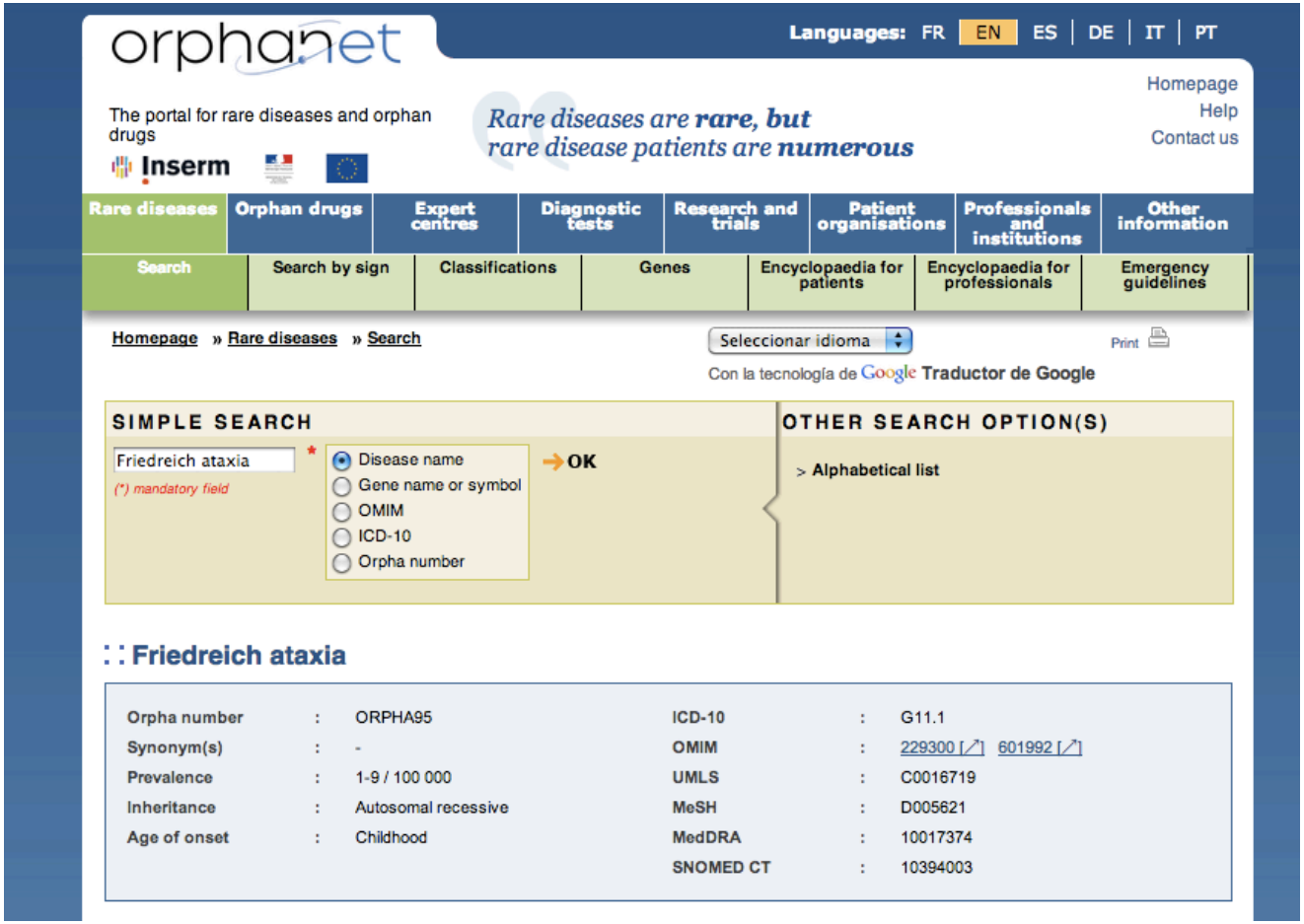
- OMIM
  - http://www.omim.org/
  - Search by disease name:
    - Ej: Friedreich ataxia



PhenUMA Input→ 1. Input Options → Identifiers for Diseases

Identifiers for Diseases: Orphanum

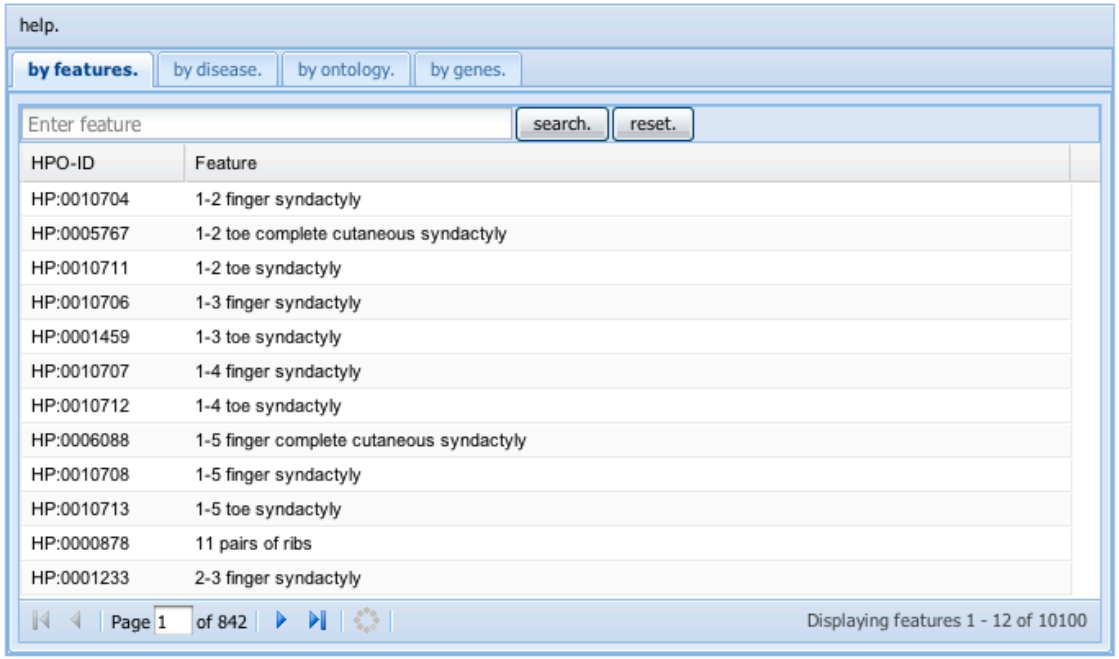
- Rare diesases



PhenUMA Input→ 1. Input Options → Identifiers for Diseases

Phenotypes

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



PhenUMA Input→ 1. Input Options → Identifiers for Diseases