

Topic 8. Phenotypes and diseases

The drug discovery timeline. OMIM. Open Targets. InterMine.

The Genotype to Phenotype challenge

Topic 5 – Genes & Genomes

Topic 6 – Functional genomics

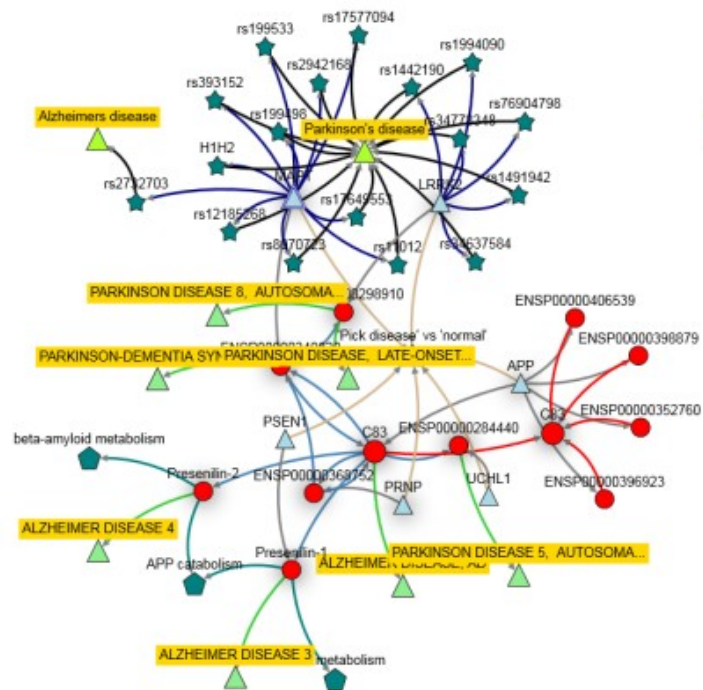
Topic 7 – Networks & Pathways

Topic 8 – Phenotypes & Diseases



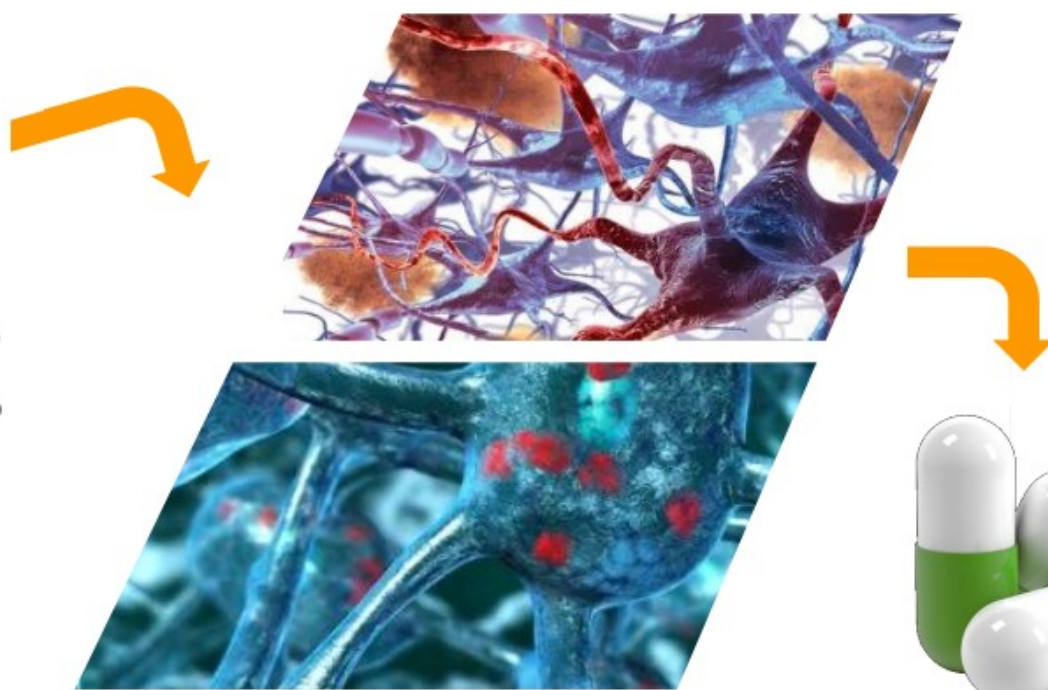
Genotype

GWAS, QTL



Biological Knowledge Discovery

Data selection, processing, transformation,
integration, interpretation



Phenotype

Alzheimer, Parkinson

Drugs

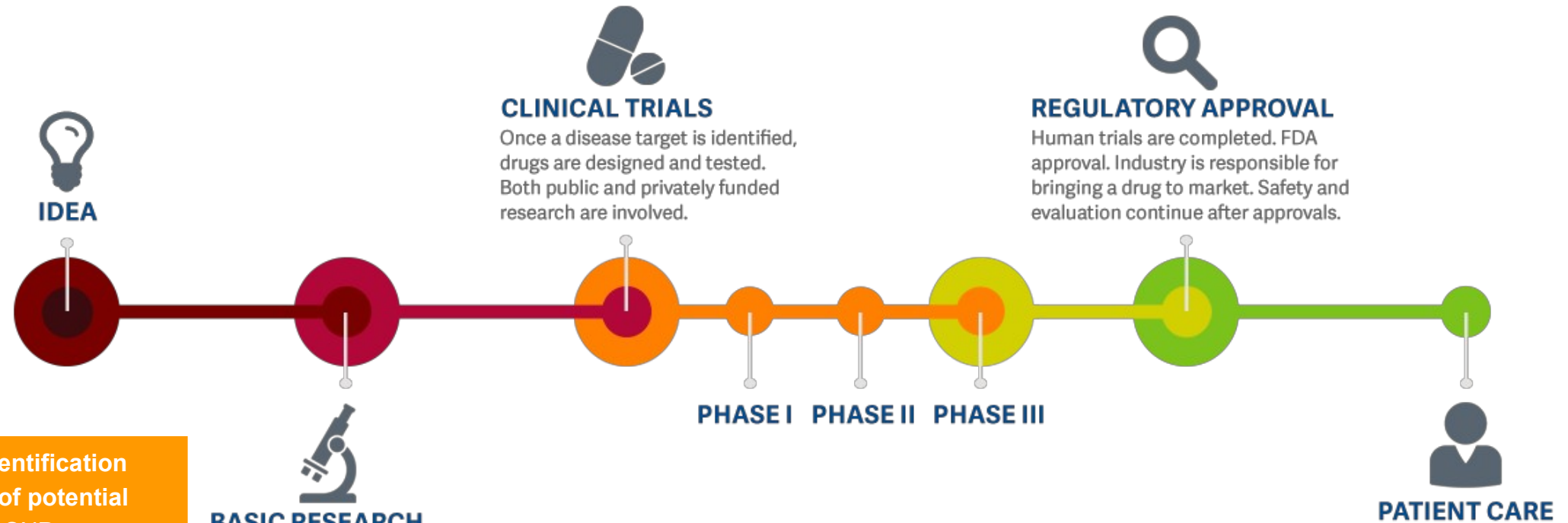
Precision medicine

Drug discovery timeline

1. DISCOVERY

2. DEVELOPMENT

3. DELIVERY



Improve the identification and selection of potential targets e.g. SNPs, gene expression, DNA/RNA-protein interactions, etc.

Characterization of the disease process and identification of drug targets

Less than 10% of candidate targets turn out to be valid

De novo drug discovery and development is a 10-17 year process

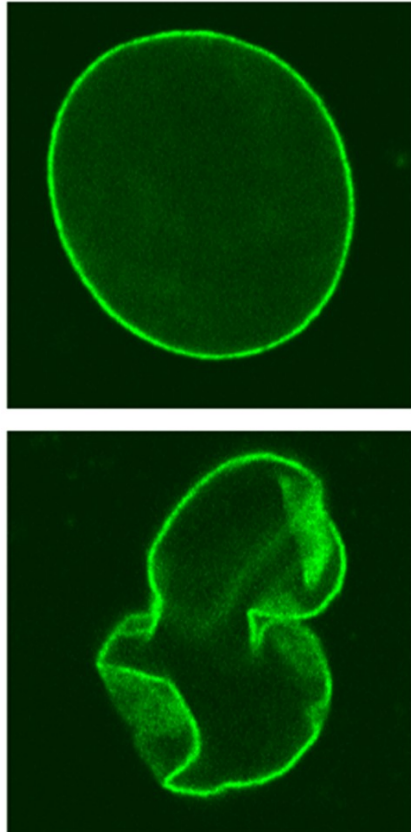
Where to start?

We are looking for better understanding of Hutchinson-Gilford progeria syndrome

Yes, and ultimately, interested in identifying drug targets for this extremely rare disease

Where could we start finding more about this disease and possible targets?

Where to start?



Google

Hutchinson-Gilford progeria

Totes Imatges Vídeos Maps Més Configuració Eines

Aproximadament 132.000 resultats (0,66 segons)

Consell: Cerca només resultats en **català** . Podeu especificar l'idioma de la cerca a **Preferències**

Articles acadèmics per Hutchinson-Gilford progeria

Lamin a truncation in **Hutchinson-Gilford progeria** - De **Sandre-Giovannoli** - Citat per 972

... in lamin A cause **Hutchinson-Gilford progeria** ... - **Eriksson** - Citat per 1499

... in nuclear architecture in **Hutchinson-Gilford progeria** ... - **Goldman** - Citat per 758

Hutchinson-Gilford progeria syndrome - Genetics Home Reference

<https://ghr.nlm.nih.gov/.../hutchinson-gilford-progeria-syndr...> ▼ Tradueix aquesta pàgina

Hutchinson-Gilford progeria syndrome is a genetic condition characterized by the dramatic, rapid appearance of aging beginning in childhood. Affected children ...

Progeria - Wikipedia

<https://en.wikipedia.org/wiki/Progeria> ▼ Tradueix aquesta pàgina

Progeria is an extremely rare genetic disorder in which symptoms resembling aspects of aging ... The condition was later named **Hutchinson-Gilford progeria syndrome** . The word progeria comes from the Greek words "pro" (πρό), meaning ...

Sam Berns · **Hallermann-Streiff syndrome** · **Progeria Research Foundation**

Where to start?

NCBI Resources [x] How To [x] Sign in to NCBI

PubMed.gov US National Library of Medicine National Institutes of Health

PubMed [x] Hutchinson-Gilford progeria [x] Search

Create RSS Create alert Advanced Help

Article types
Clinical Trial
Review
Customize ...

Text availability
Abstract
Free full text
Full text

PubMed Commons
Reader comments
Trending articles

Publication dates
5 years
10 years
Custom range...

Species
Humans
Other Animals

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Format: Summary ▾ Sort by: Most Recent ▾ Per page: 20 ▾ Send to ▾ Filters: [Manage Filters](#)

Best matches for Hutchinson-Gilford progeria:

[Hutchinson-Gilford progeria syndrome.](#)
Ullrich NJ et al. Handb Clin Neurol. (2015)

[Epigenetic involvement in Hutchinson-Gilford progeria syndrome: a mini-review.](#)
Arancio W et al. Gerontology. (2014)

[Hutchinson-Gilford progeria syndrome through the lens of transcription.](#)
Prokocimer M et al. Aging Cell. (2013)

[Switch to our new best match sort order](#)

Results by year



Download CSV

Related searches

[hutchinson-gilford progeria syndrome](#)

PMC Images search for Hutchinson-Gilford progeria



Search results

Items: 1 to 20 of 610 << First < Prev Page 1 of 31 Next > Last >>

1. [The Potential of iPSCs for the Treatment of Premature Aging Disorders.](#)
Compagnucci C, Bertini E.
Int J Mol Sci. 2017 Nov 7;18(11). pii: E2350. doi: 10.3390/ijms18112350. Review.
PMID: 29112121 [Free Article](#)
[Similar articles](#)

2. [Protein sequestration at the nuclear periphery as a potential regulatory mechanism in premature aging.](#)
Serebryanny L, Misteli T.
J Cell Biol. 2017 Oct 19. pii: jcb.201706061. doi: 10.1083/jcb.201706061. [Epub ahead of print] Review.
PMID: 29051264

Where to start?

NCBI

Resources

How To

Sign in to NCBI

OMIM


OMIM

Hutchinson-Gilford progeria

Search

Create alert Limits Advanced

Help



OMIM

OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org.

[Manage Filters](#)

☐ [#176670 - HUTCHINSON-GILFORD PROGERIA SYNDROME; HGPS](#)

1. **PROGERIA SYNDROME, CHILDHOOD-ONSET, INCLUDED**
OMIM: 176670
[Gene summaries](#) [Genetic tests](#) [Medical literature](#)

☐ [*150330 - LAMIN A/C; LMNA](#)

2. **LAMIN A, INCLUDED**
Cytogenetic locations: 1q22
OMIM: 150330
[Gene summaries](#) [Genetic tests](#) [Medical literature](#)

☐ [#248370 - MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA](#)

3. **MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY, ATYPICAL, INCLUDED**
Cytogenetic locations: 1q22
OMIM: 248370
[Gene summaries](#) [Genetic tests](#) [Medical literature](#)

☐ [*606480 - ZINC METALLOPROTEINASE STE24; ZMPSTE24](#)

4. **Cytogenetic locations: 1p34.2**
OMIM: 606480
[Gene summaries](#) [Genetic tests](#) [Medical literature](#)

Find related data

Database: Select

Find items

Search details

Hutchinson-Gilford[All Fields] AND progeria[All Fields]

Search

See more...

Recent activity

Turn Off Clear

Hutchinson-Gilford progeria (13)

Integration, integration, integration

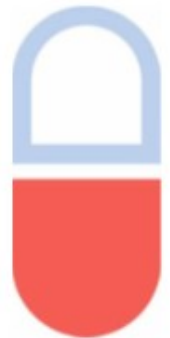
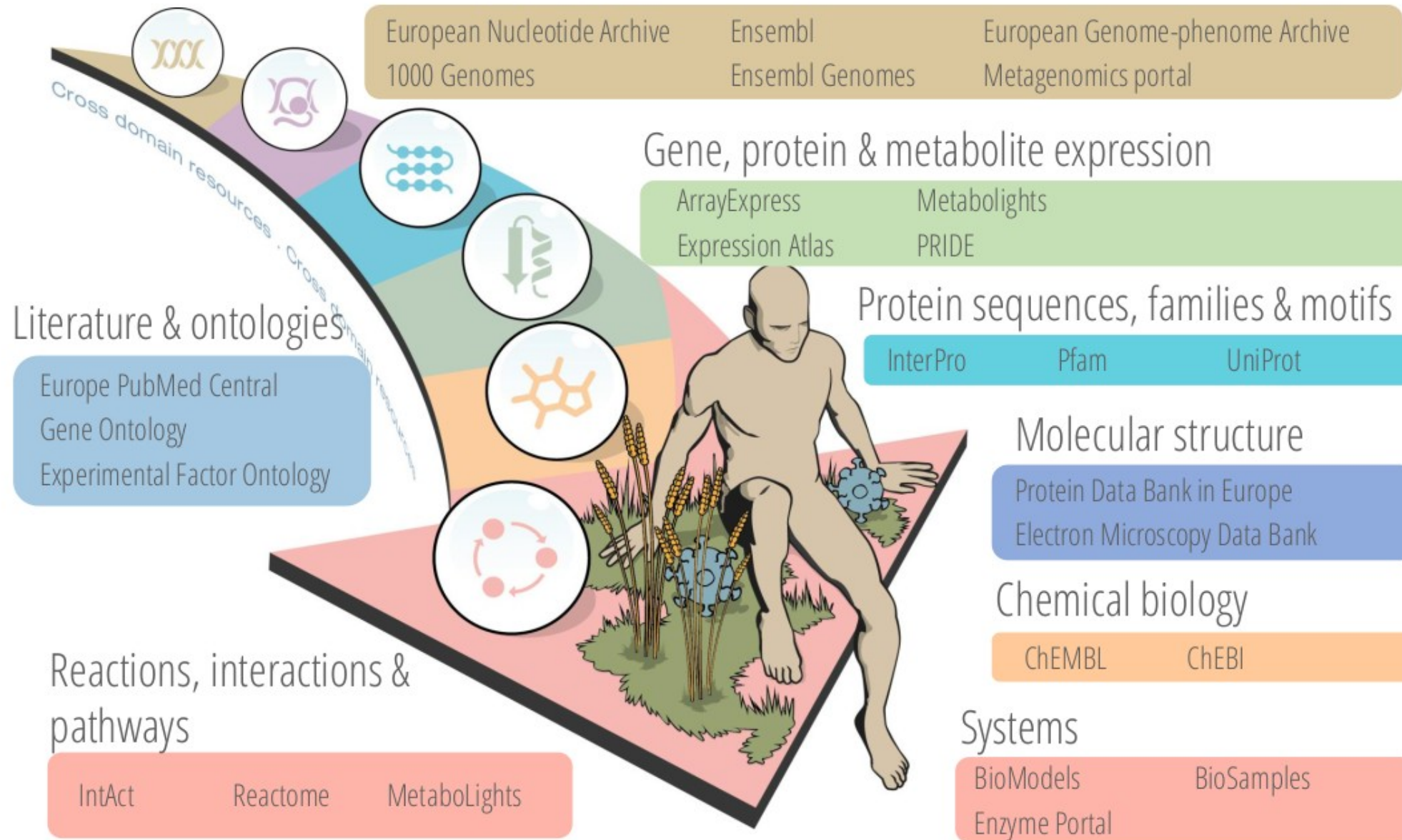
A background image showing three scientists in a laboratory setting. A woman with glasses and a ponytail is in the foreground, looking down. Two other scientists are visible behind her, also looking down. They are all wearing white lab coats. The background is slightly blurred, showing lab equipment and shelves.

I wish I could go to one place only and get as much information in an easy fashion

Yes, one one-stop shop with comprehensive data that I can trust and rank, with intuitive visualization

That'd be fab! It'd be much quicker to carry out my experiments in the lab identifying and prioritizing new targets

Public databases for drug discovery



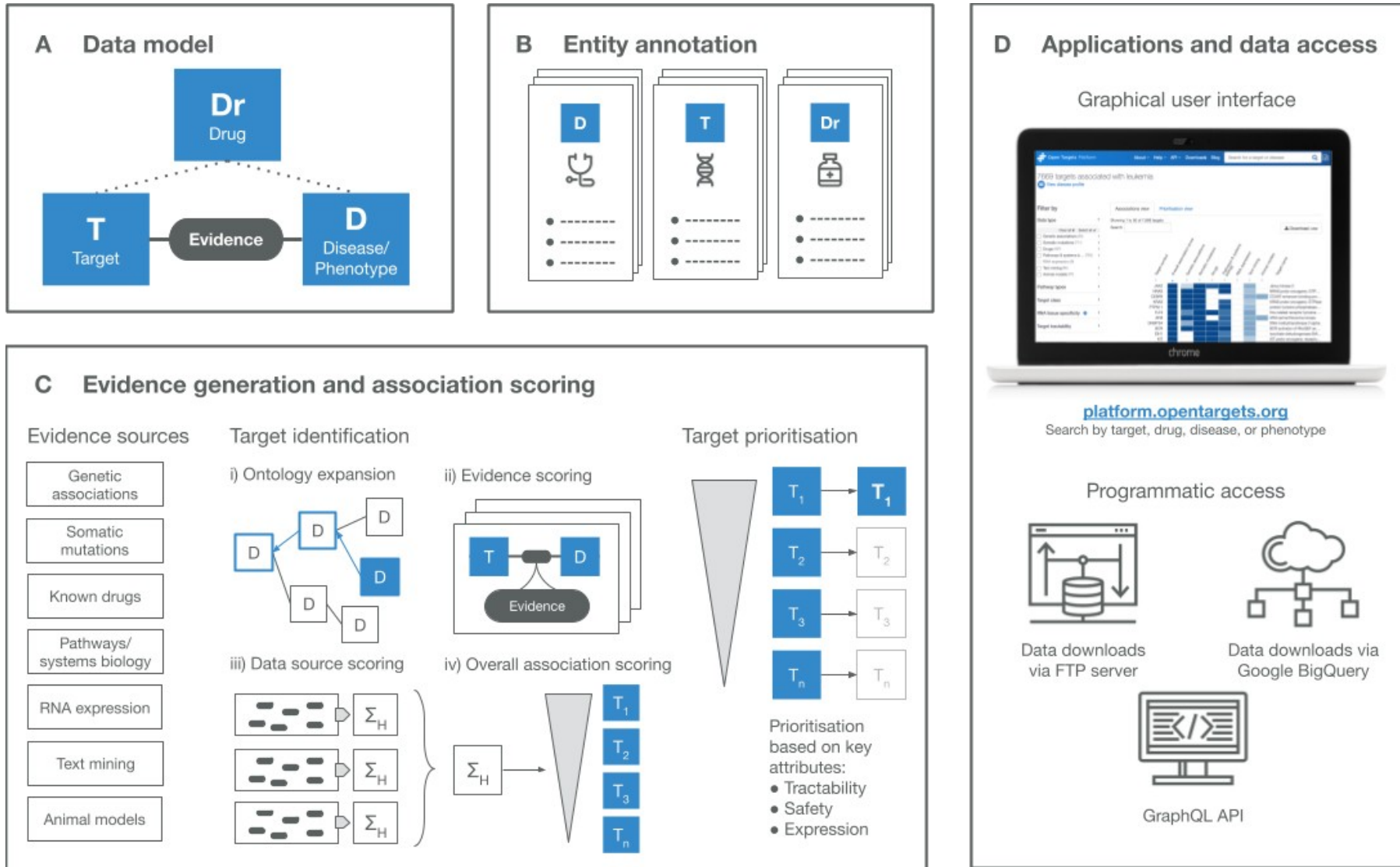
Open Targets



Open Targets is an innovative, large-scale, multi-year, public-private partnership that uses human genetics and genomics data for systematic drug target identification and prioritization.

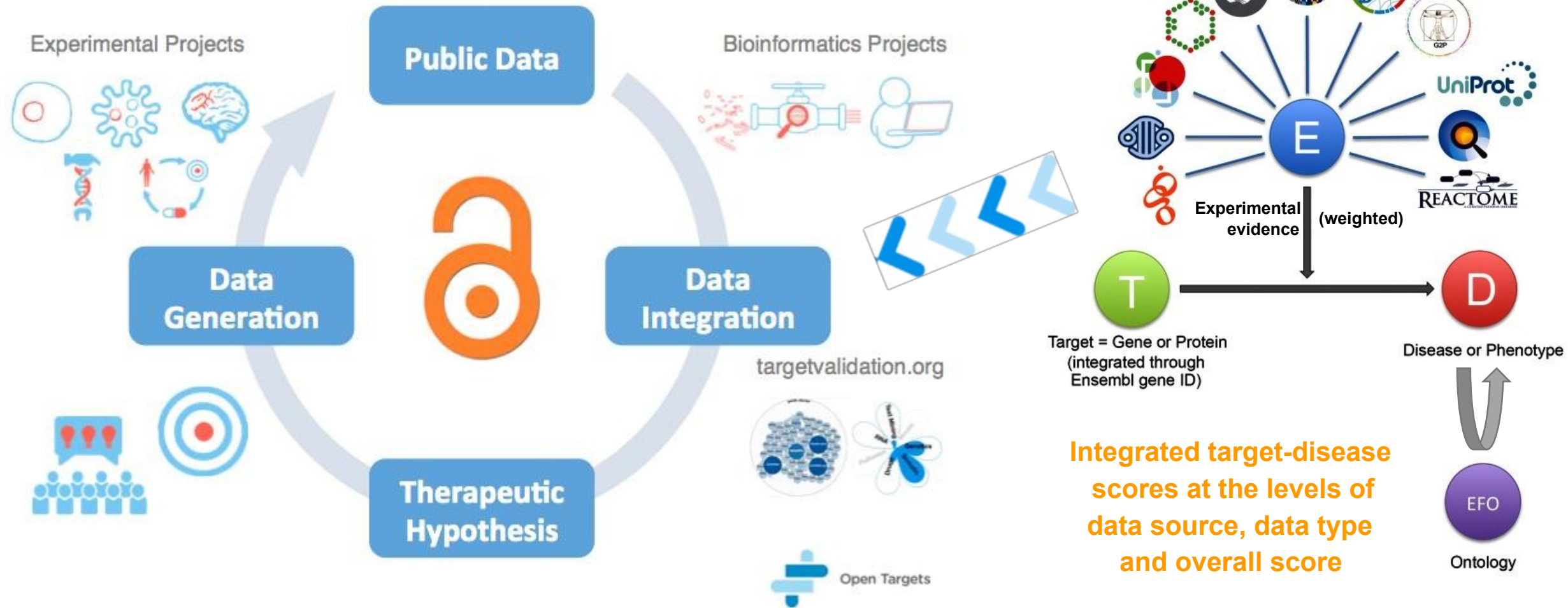
<https://platform.opentargets.org/>

Open Targets



<https://platform-docs.opentargets.org/getting-started>

Open Targets



Open Targets: sources of data

Genome Wide Association Studies

<https://www.ebi.ac.uk/gwas>

PhenoDigm: association of mouse models with disease

<http://www.sanger.ac.uk/science/tools/phenodigm>

ChEMBL: known drugs linked to a disease
and a known target

<https://www.ebi.ac.uk/chembl>

Europe PMC: mining titles, abstracts, full text

<https://europepmc.org>

COSMIC: Catalog Of Somatic Mutations In
Cancer

<http://cancer.sanger.ac.uk/cosmic/census>

IntOGen: Integrative Onco Genomics

<http://www.intogen.org>

European Variation Archive: germline and somatic variants

<https://www.ebi.ac.uk/eva>

Gene2Phenotype: variants, genes, phenotypes in
developmental disorders

<https://www.ebi.ac.uk/gene2phenotype>

Protein: sequence, annotation, function

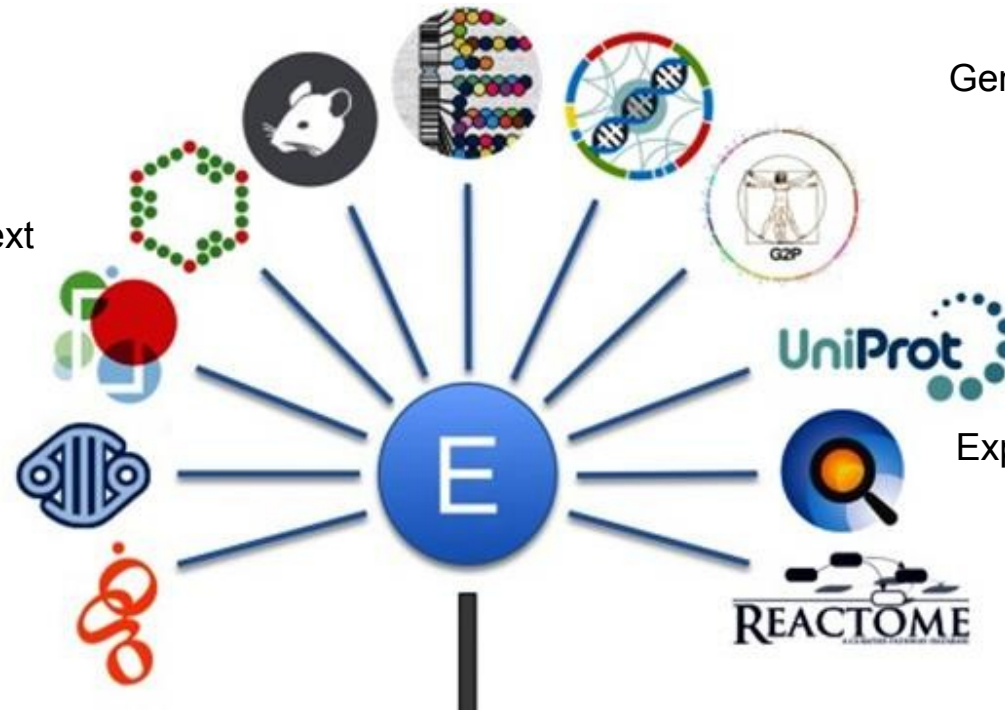
<https://www.ebi.ac.uk/uniprot>

Expression Atlas: baseline/differential expression

<https://www.ebi.ac.uk/gxa>

Biochemical reactions and pathways

<https://reactome.org>



Open Targets: examples of questions

Which targets are associated with a disease?

What evidence supports this target-disease association?

Are there FDA-approved drugs for this association?

For a target, are there other diseases associated with it?

If so, can I get associations for diseases from different therapeutic areas?

What else can I find out about my drug target?


Can I find out about the mechanisms of the disease?



Bachelor's Degree in Bioinformatics

Open Targets Use Case: Hutchinson-Gilford progeria syndrome

- O
- KD
- CS
- B



Hutchinson-Gilford progeria syndrome
EFO: Orphanet_740 | MESH: D000593 | OMIM: 103200 | NCIt: C34951 | ICD10: E34.8 | MONDO: 0008310

Associated targets

Profile

Information about the disease

Description

Hutchinson-Gilford progeria syndrome is a rare, fatal, autosomal dominant and premature aging disease, beginning in childhood and characterized by growth reduction, failure to thrive, a typical facial appearance (prominent forehead, protuberant eyes, thin nose with a beaked tip, thin lips, micrognathia and protruding ears) and distinct dermatologic features ... [show more]

Synonyms

progeria Progeria HGPS premature senility syndrome Hutchinson-Gilford disease
Hutchinson-Gilford progeria syndrome Hutchinson Gilford syndrome Hutchinson Gilford progeria syndrome
progeria syndrome, childhood-onset

Ontology

Belongs to 3 therapeutic areas

Known Drugs

3 drugs with 4 targets

Clinical signs and symptoms

104 phenotypes

Bibliography

1.692 publications

Ontology

Ontology subgraph including children, ancestors and therapeutic areas of **Hutchinson-Gilford progeria syndrome**. Source: EFO.

therapeutic area

disease

descendants

ancestors

Hutchinson-Gilford progeria syndrome

GENERAL

SPECIFIC

musculoskeletal or connective tissue disease

disease of visual system

genetic, familial or congenital

musculoskeletal system disease

connective tissue disease

eye disease

congenital abnormality

genetic disorder

hereditary connective tissue disease

Rare genetic eye disease

developmental defect during embryonic development

Rare genetic developmental disease

autosomal genetic disease

laminopathy

bone disease

Rare genetic bone developmental disease

Ectodermal malformation syndrome

progeroid syndrome

Malformation syndrome with progeroid features

Genetic progeroid syndrome

autosomal recessive disease

bone development disease

Rare genetic bone disease

Primary bone dysplasia

Primary osteolysis

Hutchinson-Gilford progeria syndrome

Known Drugs

Public Databases in Health and Life Sciences

Irepan Salvador, UAB

Bachelor's Degree in Bioinformatics

Open Targets Use Case: Hutchinson-Gilford progeria syndrome

- Home
- O
- KD
- CS
- B

KD

Known Drugs

Clinical precedence for investigational or approved drugs indicated for **Hutchinson-Gilford progeria syndrome** and curated mechanism of action. Source: [ChEMBL](#).

Q Search

Download table as

JSON

CSV

TSV

Disease information		Drug information			Target information		Clinical trials information		
Disease	Drug	Type	Mechanism Of Action	Action Type	Symbol	Name	Phase	Status	Source
Hutchinson-Gilford progeria syndrome	LONAFARNIB	Small molecule	Protein farnesyltransferase inhibitor	Inhibitor	FNTB	farnesyltransferase, CAAX box, beta	Phase IV	N/A	FDA
Hutchinson-Gilford progeria syndrome	LONAFARNIB	Small molecule	Protein farnesyltransferase inhibitor	Inhibitor	FNTA	farnesyltransferase, CAAX box, alpha	Phase IV	N/A	FDA
Hutchinson-Gilford progeria syndrome	PRAVASTATIN	Small molecule	HMG-CoA reductase inhibitor	Inhibitor	HMGCR	3-hydroxy-3-methylglutaryl-CoA reductase	Phase II	Completed	2 references
Hutchinson-Gilford progeria syndrome	ZOLEDRONIC ACID	Small molecule	Farnesyl diphosphate synthase inhibitor	Inhibitor	FDPS	farnesyl diphosphate synthase	Phase II	Completed	2 references
Hutchinson-Gilford progeria syndrome	LONAFARNIB	Small molecule	Protein farnesyltransferase inhibitor	Inhibitor	FNTA	farnesyltransferase, CAAX box, alpha	Phase II	Completed	2 references
Hutchinson-Gilford progeria syndrome	LONAFARNIB	Small molecule	Protein farnesyltransferase inhibitor	Inhibitor	FNTB	farnesyltransferase, CAAX box, beta	Phase II	Completed	2 references

Rows per page: 10 1-6 of 6 < >

CS

Clinical signs and symptoms

Clinical signs and symptoms observed in **Hutchinson-Gilford progeria syndrome**. Source: [EFO](#), [MONDO](#), [HPO](#).

Q Search

Download table as

JSON

CSV

TSV

Qualifier	Phenotype	Phenotype ID	Aspect	Frequency	Onset	Modifier	Sex	Evidence	Source	References
	Relative macrocephaly?	HP:0004482	p?	Frequent	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Hip pain?	HP:0030838	p?	Occasional	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Short lingual frenulum?	HP:0000200	p?	Frequent	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Limitation of movement at ankles?	HP:0010505	p?	Occasional	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Stroke?	HP:0001297	p?	Occasional	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Ankyloglossia?	HP:0010296	p?	Frequent	N/A	N/A	N/A	TAS?	HPO	ORPHA:740
	Delayed menarche?	HP:0012569	p?	Frequent	N/A	N/A	N/A	TAS?	HPO	ORPHA:740

Open Targets Use Case: Hutchinson-Gilford progeria syndrome



Back to the targets...

Hutchinson-Gilford progeria syndrome

EFO: Orphanet: 128001 | UMLS: CN236401, C0033300 | NCit: C34951 | ICD10: E34.8 | MONDO: 0008310

Associated targets

Profile

452 targets associated with Hutchinson-Gilford progeria syndrome

Filter by

Evidence-specific filters

Data Types

Target-specific filters

- Pathway Types
- Target Classes
- Tractability Antibody
- Tractability PROTAC
- Tractability Small Molecule

Search


Download table as JSON CSV TSV

Symbol	Overall association score	Genetic associations	Somatic mutations	Drugs	Pathways & systems biology	Text mining	RNA expression	Animal models	Target name
LMNA									lamin A/C
FNTA									farnesyltransferase, CAAX box, a...
FNTB									farnesyltransferase, CAAX box, ...
ZMPSTE24									zinc metalloproteinase STE24
ERCC4									ERCC excision repair 4, endonuc...
FDPS									farnesyl diphosphate synthase
HMGCR									3-hydroxy-3-methylglutaryl-CoA...
ATM									ATM serine/threonine kinase
HDAC2									histone deacetylase 2
CDKN1A									cyclin dependent kinase inhibitor...
PML									PML nuclear body scaffold
VDR									vitamin D receptor
XPO1									exportin 1
RB1									RB transcriptional corepressor 1
BMP4									bone morphogenetic protein 4
CAT									catalase
NPY									neuropeptide Y
MMP13									matrix metalloproteinase 13

Open Targets Use Case: Hutchinson-Gilford progeria syndrome

- OG
- PC
- CV
- GE
- GP
- UL
- UV
- CG
- ON
- CC
- IO
- CS
- CE
- PS
- SE
- PY


Evidence for LMNA in Hutchinson-Gilford progeria syndrome



LMNA

Description
Lamins are components of the nuclear lamina, a fibrous layer on the nucleoplasmic side of the inner nuclear membrane, which is thought to provide a framework for the nuclear envelope and may also interact with chromatin. Lamin A and C are present in equal amounts in the lamina of mammals. Recruited by DNA repair proteins XRCC4 and IFFO1 to the DNA ... [show more]






















Synonyms
Prelamin-A/C Lamin-A/C 70 kDa lamin Renal carcinoma antigen NY-REN-32 LMNA LMN1 HGPS MADA mandibuloacral dysplasia type A CDCD1 ... [show more]



Hutchinson-Gilford progeria syndrome

Description
Hutchinson-Gilford progeria syndrome is a rare, fatal, autosomal dominant and premature aging disease, beginning in childhood and characterized by growth reduction, failure to thrive, a typical facial appearance (prominent forehead, protuberant eyes, thin nose with a beaked tip, thin lips, micrognathia and protruding ears) and distinct dermatologic ... [show more]


Synonyms
progeria Progeria HGPS premature senility syndrome Hutchinson-Gilford disease Hutchinson-Gilford progeria syndrome Hutchinson Gilford syndrome Hutchinson Gilford progeria syndrome progeria syndrome, childhood-onset

<div>OG OT Genetics Portal Genetic associations</div> <div>no data</div>	<div>PC PheWAS Catalog Genetic associations</div> <div>no data</div>	<div>CV ClinVar Genetic associations</div> <div>93 entries</div>	<div>GE GEL PanelApp Genetic associations</div> <div>6 entries</div>	<div>GP Gene2Phenotype Genetic associations</div> <div>2 entries</div>	<div>UL UniProt literature Genetic associations</div> <div>1 entry</div>
<div>UV UniProt variants Genetic associations</div> <div>10 entries</div>	<div>CG ClinGen Genetic associations</div> <div>no data</div>	<div>ON Orphanet Genetic associations</div> <div>1 entry</div>	<div>CC Cancer Gene Census Somatic mutations</div> <div>no data</div>	<div>IO IntOGen Somatic mutations</div> <div>no data</div>	<div>CS ClinVar (somatic) Somatic mutations</div> <div>no data</div>
<div>CE ChEMBL Drugs</div> <div>no data</div>	<div>PS Project Score Pathways & systems biology</div> <div>no data</div>	<div>SE SLAPenrich Pathways & systems biology</div> <div>no data</div>	<div>PY PROGENy Pathways & systems biology</div> <div>no data</div>	<div>RT Reactome Pathways & systems biology</div> <div>no data</div>	<div>GS Gene signatures Pathways & systems biology</div> <div></div>
<div>EP Europe PMC Text mining</div> <div></div>	<div>EA Expression Atlas RNA expression</div> <div></div>	<div>PH PhenoDigm Animal models</div> <div></div>			

Details of all evidences supporting this association

Open Targets Use Case: Hutchinson-Gilford progeria syndrome

- KD
- TR
- S
- CP
- BE
- GO
- GC
- PV
- MI
- PW
- CH
- MP
- CG
- SL
- B

 **LMNA** lamin A/C

Ensembl: [ENSG00000160789](#) | UniProt: [P02545](#) | GeneCards: [LMNA](#) | HGNC: [LMNA](#) | Project Score[®]: [SIDG16660](#)

Associated diseases

Profile

Description

Lamins are components of the nuclear lamina, a fibrous layer on the nucleoplasmic side of the inner nuclear membrane, which is thought to provide a framework for the nuclear envelope and may also interact with chromatin. Lamin A and C are present in equal amounts in the lamina of mammals. Recruited by DNA repair proteins XRCC4 and IFFO1 to the DNA double-
... [show more]

Synonyms

HGPS

MADA

mandibuloacral dysplasia type A

Prelamin-A/C

Lamin-A/C

70 kDa lamin

Renal carcinoma antigen NY-REN-32

LMNA

LMN1

CDCD1

... [show more]

<div><div>KD</div><div>Known Drugs</div></div> <div>no data</div>	<div><div>TR</div><div>Tractability</div></div> <div>Assessment available</div>	<div><div>S</div><div>Safety</div></div> <div>no data</div>	<div><div>CP</div><div>Chemical Probes</div></div> <div>no data</div>	<div><div>BE</div><div>Baseline Expression</div></div> <div>RNA • Protein</div>	<div><div>GO</div><div>Gene Ontology</div></div> <div>72 terms in total 31 MF • 18 BP • 23 CC</div>
<div><div>GC</div><div>Genetic Constraint</div></div> <div>very high constraint</div>	<div><div>PV</div><div>ProtVista</div></div> <div>Positional, Structural and Functional Information</div>	<div><div>MI</div><div>Molecular Interactions</div></div> <div>2397 physical or functional interactors</div>	<div><div>PW</div><div>Pathways</div></div> <div>8 Reactome pathways</div>	<div><div>CH</div><div>Cancer Hallmarks</div></div> <div>1 hallmarks 0 promoted • 1 suppressed</div>	<div><div>MP</div><div>Mouse Phenotypes</div></div> <div>241 distinct phenotypes</div>
<div><div>CG</div><div>Comparative Genomics</div></div> <div>10 orthologues and 66 paralogues</div>	<div><div>SL</div><div>Subcellular Location</div></div> <div>8 subcellular locations</div>	<div><div>B</div><div>Bibliography</div></div> <div>8,896 publications</div>			

TR

Tractability

Target tractability assessment for **LMNA**. Source: [Open Targets](#).

<div>Small molecule</div> <div><div>✕</div> Approved Drug</div> <div><div>✕</div> Advanced Clinical</div> <div><div>✕</div> Phase 1 Clinical</div>
--

 Antibody ✕ Approved Drug ✕ Advanced Clinical ✕ Phase 1 Clinical |

Open Targets Use Case: Hutchinson-Gilford progeria syndrome

Diseases are grouped into bubbles based on the disease ontology

[View LMNA in Open Targets Genetics](#)

02545 | GeneCards: LMNA | HGNC: LMNA | Project Score: SIDG16660

943 diseases or phenotypes associated with LMNA

Filter by

Evidence-specific filters

Data Types

Disease/phenotype-specific filters

Therapeutic Areas

Table Bubbles Graph

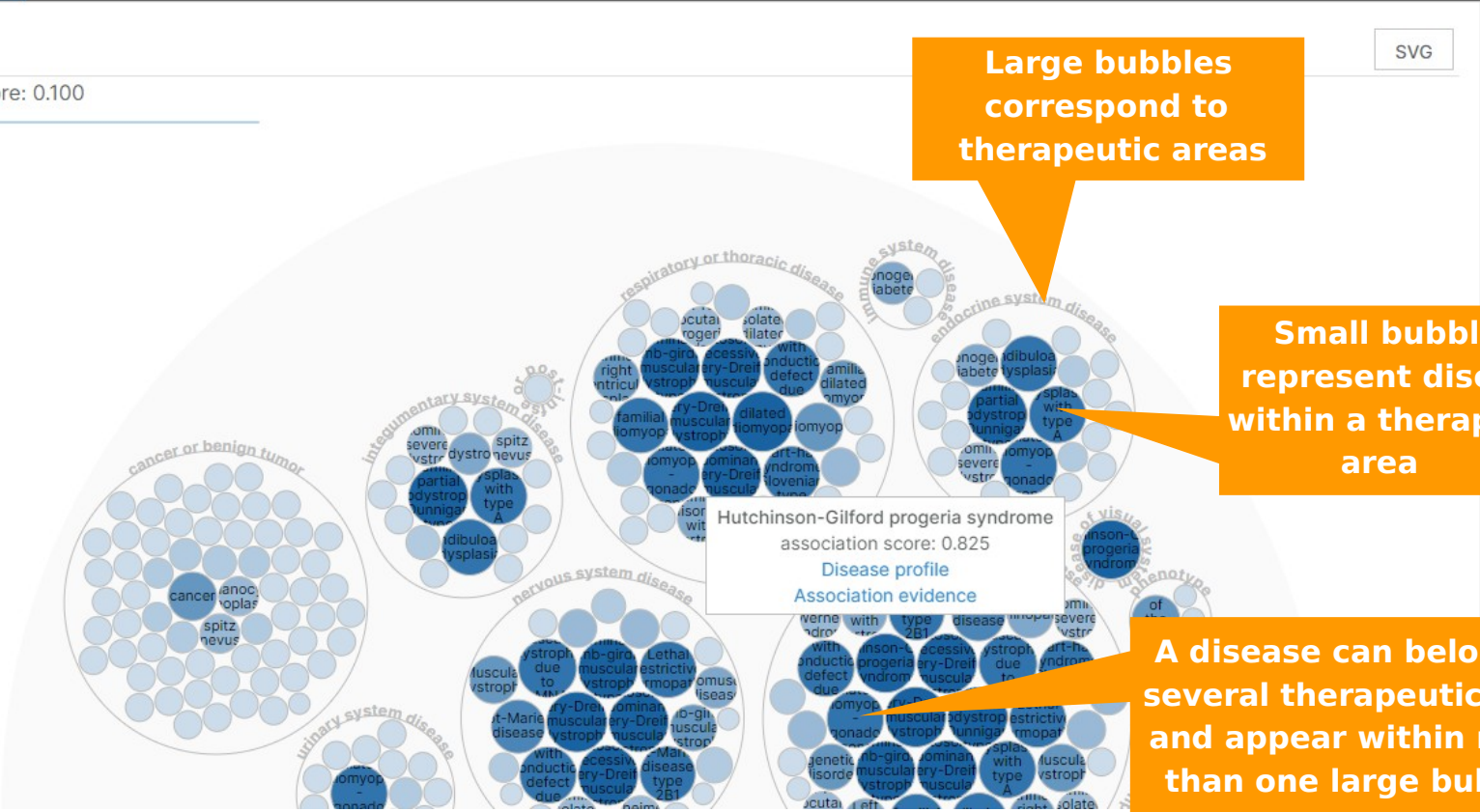
Minimum Score: 0.100

SVG

Large bubbles correspond to therapeutic areas

Small bubbles represent diseases within a therapeutic area

Strength of the association between target and disease



A disease can belong to several therapeutic areas and appear within more than one large bubble

Disparate data in, unified data out

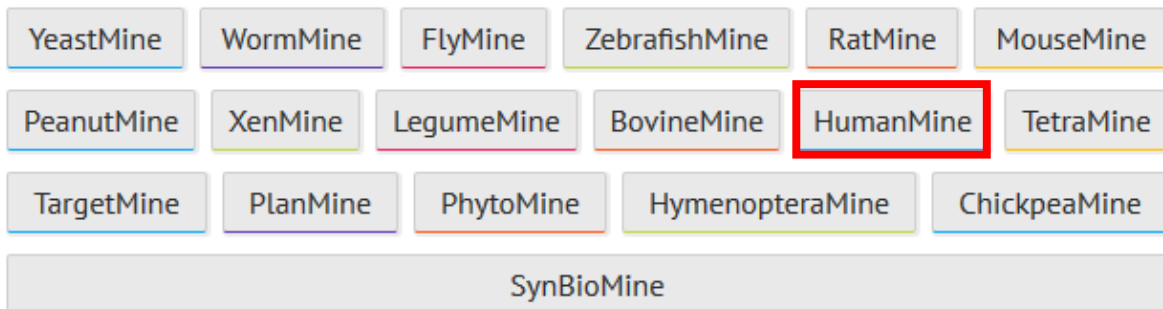
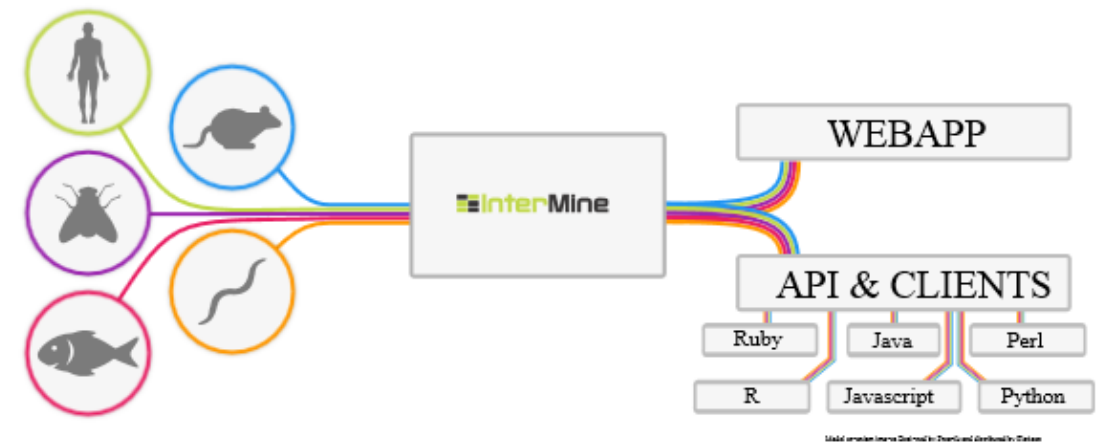
InterMine integrates biological data sources, making it easy to query and analyse data.

InterMine is [open source](#) (LGPL 2.1) and free to use.

It's a software system that you can [install on your own servers](#) to make data available on the web.

There are many different InterMines worldwide, covering a broad range of model organisms and life science research areas.

<http://intermine.org/>



There's an InterMine for (almost) anyone

With InterMine you can explore organism and other research data provided by many different organizations, moving between databases using criteria such as homology.

This is a sample of the InterMine installations available - click on one to go straight to that service.

InterMine: Querying the HumanMine

EX BLUEGENES

HomeUploadListsTemplatesRegionsQuery Builder

Search for any term

HUMANMINE

LOGIN

Search by names, identifiers or keywords for genes, proteins, pathways, ontology terms, authors, etc.

Lists of identifiers; e.g. gene or SNP identifiers

Predefined queries; e.g. "GO term Genes" or "Gene Tissue Expression"

Search features that overlap a list of genome coordinates

Flexible query interface to view the data model, apply constraints and select output

HumanMine v11 November 2011

Integrated database of

HumanMine. Please check out our new user documentation. access the old interface by clicking here.

Search for any term

Genes, proteins, pathways, ontology terms, authors, etc.

Go by Most Popular Queries

FUNCTION

EXPRESSION

DISEASE

PROTEINS

GENOMICS

INTERACTIONS

Gene

GO term

→


→

Pathway

Genes

Web Service API

InterMine Use Case: Hutchinson-Gilford progeria (search by keyword)



Home

Upload


Lists

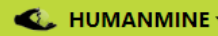
Templates

Regions

Query Builder

LMNA





LOGIN

LMNA

SEARCH

765 results for 'LMNA'

Search by keyword

Gene	Organism	<i>Rattus norvegicus</i>
	Symbol:	Lmna
	Identifiers:	RGD:620456
Gene	Organism	<i>Homo sapiens</i>
	Symbol:	LMNA
	Identifiers:	4000,ENSG00000160789
Gene	Organism	<i>Mus musculus</i>
	Symbol:	Lmna
	Identifiers:	MGI:96794
Protein	Organism:	<i>Homo sapiens</i>
	Accession:	Q8N519
	Identifiers:	Q8N519_HUMAN
Gene	Organism	<i>Homo sapiens</i>
	Symbol:	MLIP
	Identifiers:	90523,ENSG00000146147
Publication	Author:	Liu Heng
	Title:	"LMNA functions as an oncogene in hepatocellular carcinoma by regulating the proliferation and migration ability."
	Journal:	J Cell Mol Med pp. 12008-12019
Publication	Author:	Perepelina K
	Title:	"Lamin A/C mutation associated with lipodystrophy influences adipogenic differentiation of stem cells through interaction with Notch signaling."
	Journal:	Biochem Cell Biol pp. 342-348
Publication	Author:	Poitelon Yannick
	Title:	"Behavioral and molecular exploration of the AR-CMT2A mouse model Lmna (R298C/R298C)."
	Journal:	Neuromolecular Med pp. 40-52

Filter by:

Active filters:

None

Category

504

Publication

219

Exon

18

MRNA

15

UniProtFeature

4

Gene

2

Disease

1

HPOTerm

1

Protein

1

InteractionExperiment

Organism

240

H. sapiens

1

R. norvegicus

1

M. musculus

InterMine Use Case: Hutchinson-Gilford progeria (search by keyword)

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LMNA

HUMANMINE

LOGIN

Topic filter

LMNA

Gene

Gene information

Summary

Summary

Brief Description	lamin A/C	Cytological Location	1q22
Id	1274877	Length	57509
Name	lamin A/C	Organism . Name	Homo sapiens
Primary Identifier	4000	Secondary Identifier	ENSG00000160789
Symbol	LMNA	Chromosome Location	1:156082573-156140081
Strand	+	Sequence Length	57509

FASTA

Quick links

Summary

Gene Ontology

GO Annotation (29)

Subject Primary Identifier	Ontology Term Identifier	Ontology Term Name
4000	GO:0005515	protein binding
4000	GO:0005634	nucleus
4000	GO:0005635	nuclear envelope
4000	GO:0005638	lamin filament
4000	GO:0005654	nucleoplasm

Links to other Mines (orthologues)

Other mines

FlyMine

HymenopteraMine

YeastMine

Data sources

clinvar

arrayexpress-atlas

hgnc

InterMine Use Case: neurodegenerative diseases (lists)

Filter by Category
(Gene) and Organism
(H. sapiens)

Filter by:

Active filters:

Gene

H. sapiens

Category

4812 Publication

213 GWASResult

133 Author

105 Disease

78 UniProtFeature

74 InteractionExperiment

49 Protein

26 GWAS

21 ProteinDomain

17 Gene

7 MeshTerm

5 Allele

2 HPOTerm

2 OntologyTermSynonym

2 Component

2 MammalianPhenotypeTerm

2 OntologyTerm

Organism

HomeUploadListsTemplatesRegionsQuery Builderparkinson OR huntington OR alzheimerHUMANMINELOGIN

parkinson OR huntington OR alzheimer

SEARCH

Create a list

15 results for 'parkinson OR huntington OR alzheimer'

<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	
		Identifiers:	109029536
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	
		Identifiers:	109461479
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	PARK7
		Identifiers:	11315,ENSG00000116288
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	PRKN
		Identifiers:	5071,ENSG00000185345
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	BPTF
		Identifiers:	2186,ENSG00000171634
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	PSEN2
		Identifiers:	5664,ENSG00000143801
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	APOE
		Identifiers:	348,ENSG00000130203
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	PSEN1
		Identifiers:	5663,ENSG00000080815
<input type="checkbox"/>	Gene	Organism	Homo sapiens
		Symbol:	PINK1
		Identifiers:	65018,ENSG00000158828

InterMine Use Case: neurodegenerative diseases (lists)

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Activity

parkinson OR huntington OR

HUMANMINE

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MANAGE FILTERS

MANAGE RELATIONSHIPS

UNDO

SAVE LIST

PYTHON

EXPORT

Showing 1 to 10 of 15 rows

Rows per page: 10

Page 1

Gene Symbol	Gene Name	Gene Primary Identifier	Gene Secondary Identifier	Gene Length	Organism Name
APOE	apolipoprotein E	348	ENSG00000130203	3598	Homo sapiens
APP	amyloid beta precursor protein	351	ENSG00000142192	290579	Homo sapiens
ATP13A2	ATPase cation transporting 13A2	23400	ENSG00000159363	26015	Homo sapiens
BPTF	bromodomain PHD finger transcription factor	2186	ENSG00000171634	158876	Homo sapiens
GATD1	glutamine amidotransferase class 1 domain containing 1	347862	ENSG00000177225	10407	Homo sapiens
GIGYF2	GRB10 interacting GYF protein 2	26058	ENSG00000204120	163275	Homo sapiens
LRRK2	leucine rich repeat kinase 2	120892	ENSG00000188906	144396	Homo sapiens
PARK7	Parkinsonism associated deglycase	11315	ENSG00000116288	23795	Homo sapiens
PINK1	PTEN induced kinase 1	65018	ENSG00000158828	18054	Homo sapiens
PRKN	parkin RBR E3 ubiquitin protein ligase	5071	ENSG00000185345	1380386	Homo sapiens

Chromosome Distribution

Actual: number of items in this list found on each chromosome.
Expected: given the total number of items on the chromosome and the number of items in this list, the number of items expected to be found on each chromosome.

Interactions

Genes (from the list or not) that interact with genes in this list. Counts may include the same interaction more than once if observed in multiple experiments.

Generate Python, Perl, Java, Ruby, JavaScript, or XML code

Max p-value

0.05

Test Correction

Holm-Bonferroni

Background population

Choose a list

Filter enrichment results

Text to filter items

Gene Ontology Enrichment (0)

Ontology: biological_process

Publication Enrichment

Pathway Enrichment (3)

DataSet: All

Item (matches)

p-value

Amyloid fiber formation (4)

6.163200e-3

Nuclear signaling by ERBB4 (3)

7.305433e-3

Signaling by ERBB4 (3)

4.210130e-2

Protein Domain Enrichment (2)

Item (matches)

p-value

Peptidase A22A, presenilin (2)

7.659308e-3

Presenilin, C-terminal (2)

7.659308e-3

Much more information below the table!!!

Public Databases in Health and Life Sciences

Irepan Salvador, UAB

Bachelor's Degree in Bioinformatics

InterMine Use Case: genes involved in neuron death (templates)

Filter templates

Templates

Filter by category: All Disease Expression Variant **Proteins** Function GWAS Pathways Gene Ontology Genomics SNPs Interactions Homology

Filter by description: Filter text...

Gene → Pathway

For a given Gene (or List of Genes) show any associated Pathway(s) (Data Source: KEGG or REACTOME). Keywords: pathways, metabolism, cascade

[View >>](#) Categories: Function Pathways

GO term → Genes

Search for Genes in a specified organism that are associated with a particular Gene Ontology (GO) annotation.

[View >>](#) Categories: Function **Gene Ontology** Genomics

Gene → GWAS hit

For a given Gene (or List of Genes) returns a list of SNPs associated with the Gene in GWAS experiments. Results can be constrained by P-value [Format Examples: 0.05 or 2.0E-6; default = 0.01] or Phenotype [Example: *diabetes* or *obesity* or try typing your keyword to bring up the autocomplete options] [Keywords: genome wide association, study, studies, disease]

[View >>](#) Categories: GWAS

Gene → GO terms.

Search for GO annotations for a particular gene (or List of Genes).

[View >>](#) Categories: Function **Gene Ontology** Genomics

Tissue → Gene Expression (Array Express)

Show expression for a gene or list of genes associated with a specific tissue(s). Optionally constrain the P-value. Data source: The Gene Expression Atlas

InterMine Use Case: genes involved in neuron death (templates)

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Search for any term

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Filter by category

AllDiseaseExpressionVariantProteinsFunctionGWASPathwaysGene OntologyGenomicsSNPsInteractionsLiteratureComparative GenomicsHomology

Filter by description

Filter text...

Gene → Pathway

For a given Gene (or List of Genes) show any associated Pathway(s) (Data Source: KEGG or REACTOME). Keywords: pathways, metabolism, cascade

View >>

Categories: FunctionPathways

GO term → Genes

Search for Genes in a specified organism that are associated with a particular Gene Ontology (GO) annotation.

Ontology Term > Name

Like

neuron death

Organism > Short Name

=

H. sapiens

Results Preview

Gene > Primary Identifier	Gene > Symbol	Gene > Name	GO Annotation > Ontology Term . Identifier	GO Annotation > Ontology Term . Name	Organism > Short Name
9529	BAG5	BAG co-chaperone 5	GO:0070997	neuron death	H. sapiens
867	CBL	Cbl proto-oncogene	GO:0070997	neuron death	H. sapiens
10570	DPYSL4	dihydropyrimidinase like 4	GO:0070997	neuron death	H. sapiens
120892	LRRK2	leucine rich repeat kinase 2	GO:0070997	neuron death	H. sapiens
10135	NAMPT	nicotinamide phosphoribosyltransferase	GO:0070997	neuron death	H. sapiens

+ 2 more results

VIEW 7 ROWS

EDIT QUERY

Close <<

Categories: FunctionGene OntologyGenomics

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InterMine Use Case: genes involved in neuron death (templates)

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EXPORT

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Rows per page: All (7)

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Gene Primary Identifier	Gene Symbol	Gene Name	Ontology Term Identifier	Ontology Term Name	Organism Short Name
9529	BAG5	BAG cochaperone 5	GO:0070997	neuron death	H. sapiens
867	CBL	Cbl proto-oncogene	GO:0070997	neuron death	H. sapiens
10570	DPYSL4	dihydropyrimidinase like 4	GO:0070997	neuron death	H. sapiens
120892	LRRK2	leucine rich repeat kinase 2	GO:0070997	neuron death	H. sapiens
10135	NAMPT	nicotinamide phosphoribosyltransferase	GO:0070997	neuron death	H. sapiens
9481	SLC25A27	solute carrier family 25 member 27	GO:0070997	neuron death	H. sapiens
6548	SLC9A1	solute carrier family 9 member A1	GO:0070997	neuron death	H. sapiens

Widgets

Chromosome Distribution

Actual: number of items in this list found on each chromosome.
Expected: given the total number of items on the chromosome and the number of items in this list, the number of items expected to be found on each chromosome.

All Genes in the table have been analysed in this widget.

Organism: Homo sapiens

Actual Expected

Interactions

Genes (from the list or not) that interact with genes in this list. Counts may include the same interaction more than once if observed in multiple experiments.

All Genes in the table have been analysed in this widget.

VIEW ALL

☐ BioEntity.primaryIdentifier BioEntity.name

☐ 3308 heat shock protein family A

Enrichment

Enrichment column: Gene

Max p-value: 0.05

Test Correction: Holm-Bonferroni

Background population: Choose a list

Filter enrichment results: Text to filter items

Gene Ontology Enrichment (0)

Ontology: biological_process

Publication Enrichment

Pathway Enrichment (0)

DataSet: All

Protein Domain Enrichment (0)

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