

# Rare Disease Alert System (RDAS)

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#AMIA2023



# Burden of Rare Diseases



## Total Economic Burden of Rare Disease in the U.S in 2019: \$966 Billion\*

### Indirect Costs & Non-medical Costs \$548 Billion

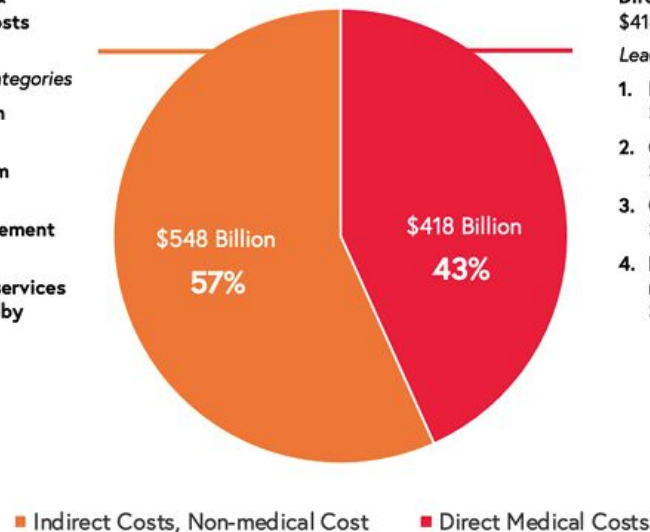
#### Leading Cost Categories

1. Absenteeism  
\$149 Billion
2. Presenteeism  
\$138 Billion
3. Forced Retirement  
\$136 Billion
4. Healthcare services not covered by insurance  
\$38 Billion

### Direct Medical Costs \$418 Billion

#### Leading Cost Categories

1. Inpatient  
\$143 Billion
2. Outpatient  
\$62 Billion
3. Other ancillary  
\$49 Billion
4. Prescription medication  
\$48 Billion



The National Economic Burden of Rare Disease Study from Everylife Foundation for Rare Diseases

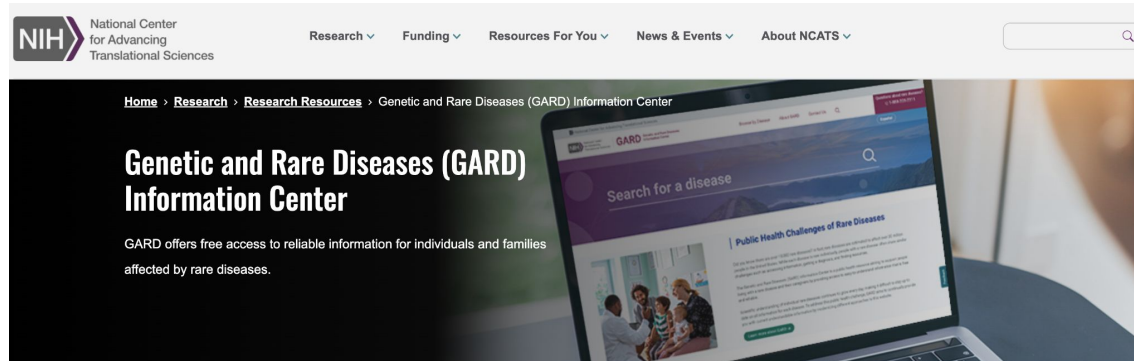
# Genetic And Rare Diseases (GARD) Program

Established in 2002

<https://rarediseases.info.nih.gov>

GARD's Mission: Provide comprehensive, plain-language information on rare diseases that is freely accessible to the public.

Currently maintain >10,000 rare diseases



# Meeting the Needs for Rare Disease Patients, Clinicians, and Scientists

LESS INFORMATION  
FEWER RESOURCES  
MORE BARRIERS TO ACCESS



**Our Goal:** To empower the research community in their rare disease research efforts and to provide an educational resource

**How?** Integrate the latest biomedical data in a structured, standardized, and semantic way

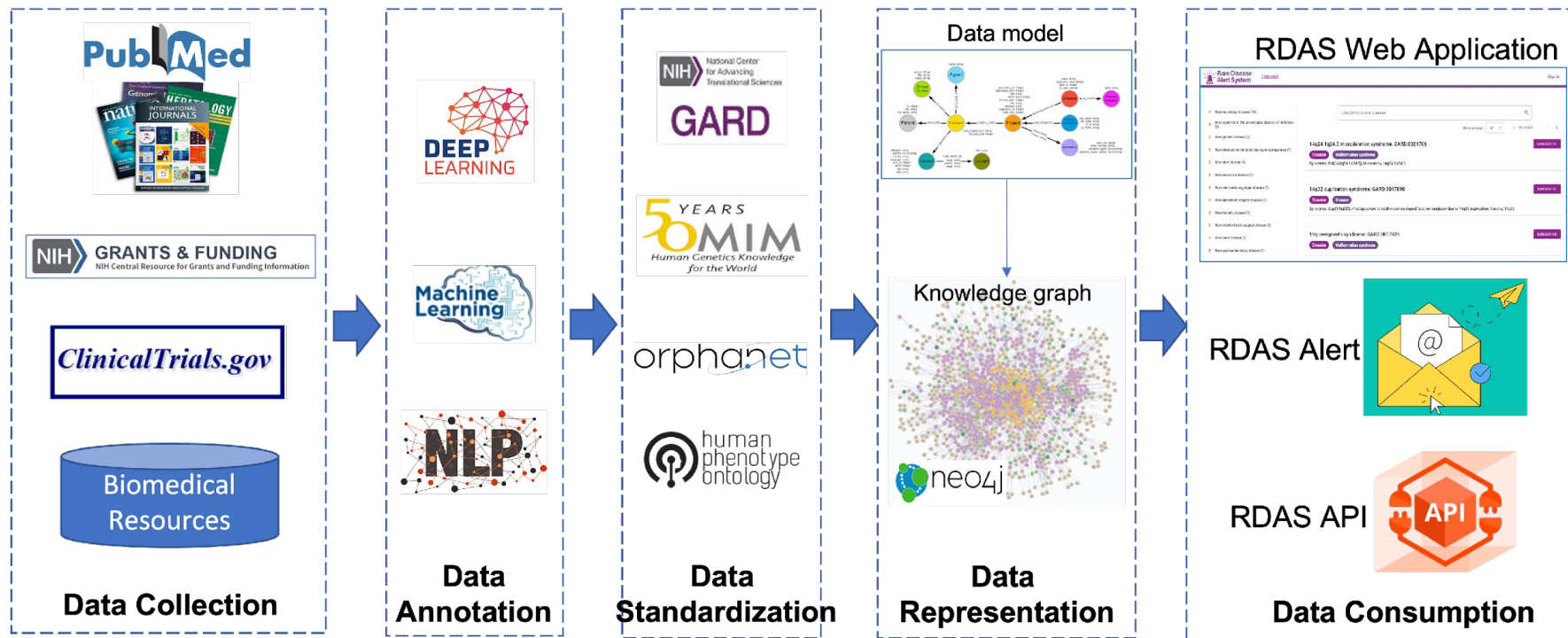


**Rare Disease  
Alert System**

## Main Features

- Integrated information from publications, clinical trials, and grants
- Diseases standardized with well-known rare disease based controlled vocabularies, GARD, Orphanet, etc.
- Timely alerts regarding updates generated for subscribers
- Ease of access via a user-friendly web interface and API

# RDAS Architecture



# RDAS neo4j – <https://rdas.ncats.nih.gov/browser>

The screenshot displays the RDAS neo4j browser interface. The browser tab shows the URL `rdas.ncats.nih.gov/browser/?iss=https%3A%2F%2Fauth.ncats.nih.gov%2F_api%2Fv2%2Fauth%2Fncats`. The left sidebar contains a menu with options: clinical, gard, grant, neo4j, **pubmed** (selected), and system. Below the menu are 'Node labels' and 'Relationship types' filters. The main area shows a Cypher query: `pubmed$ MATCH (n:Article) RETURN n LIMIT 25`. The query results are visualized as a graph with purple circular nodes and a central pie chart. The right sidebar shows 'Node properties' for an 'Article' node, including: `<id>` 552, `DateCreat` 12/08/22, `edRDAS`, `abstractText` (Very early onset Toni-Debré-Fanconi Syndrome, a disorder of proximal renal tubules of the kidney which results in the increased urinary excretion of g...), and `affiliation` (Department of Pediatric Metabolism and Nutrition, Gazi University Hospital, Ankara, Turkey.).





DISEASES APIs

Search for a rare disease



Sign In

## Meeting the Needs of Rare Disease Patients, Clinicians, and Scientists

Integrating the latest biomedical data in a structured, standardized, and semantic way to empower the rare disease research community in their efforts, and provide an educational resource to all.

Learn More



RDAS-Alert: Funded Project update regarding your subscriptions



ncatsrdas@mail.nih.gov

To: ✓ Leadman, Devon (NIH/NCATS) [C]

☺ Reply Reply All → Forward 📧 ...

Fri 11/3/2023 5:04 PM

## Rare Disease Alert System

**Timothy Sheils**

**Within the last week, 7065 new entries for your subscribed rare diseases have been added to the funded project database**

Name	GARD ID	Nodes Modified
<a href="#">Glioblastoma</a>	GARD:0002491	6878
<a href="#">Rare developmental defect during embryogenesis</a>	GARD:0022513	186
<a href="#">49, XXXXY syndrome</a>	GARD:0005679	1

Results gathered within the time period of 04/20/23-04/27/23





# RDAS APIs

## APIs

EPIDEMIOLOGY

DISEASES

PUBLICATIONS

PROJECTS

TRIALS

## EpiPipeline4RD API <sup>1.1</sup> OAS 3.0

/assets/epi-api/epi4rdas.json



National Center  
for Advancing  
Translational Sciences

GARD

Genetic and Rare Diseases  
Information Center

This API was developed by the [National Center for Advancing Translational Sciences \(NCATS\)](#) for the National Institutes of Health (NIH) [Genetic and Rare Diseases Information Center \(GARD\)](#).

It allows one to *gather* abstracts for a rare disease (query any rare disease name, synonym, or GARD ID) from two APIs, *classify those abstracts as epidemiologic*, and *extract* epidemiology information from them.

A full list of rare diseases tracked by the NIH Genetic and Rare Diseases Information Center can be found [here](#).

[Interactive User Interface](#)

[Example POST calls](#)

[GitHub Repository](#)

National Center for Advancing Translational Sciences License

Rare Disease Alert System

DISEASES APIs

SANDBOX <https://rdas.ncats.nih.gov/a> Publish

ExampleQuery x +

Documentation

Root > Query

← Query ✓

Fields ↓ ●

- ⊕ diseaseSearch(...): [GARD]
- ⊕ filterCounts(...): [FilterCount!]
- ⊕ filterCountsAggregate(...): FilterCountAggreg...
- ⊕ filterCountsConnection(...): FilterCountsConn...
- ⊕ gardLists(...): [GARDList!]
- ⊕ gardListsAggregate(...): GARDListAggregateSel...
- ⊕ gardListsConnection(...): GardListsConnection!
- ⊕ gards(...): [GARD!]

Operation

```
1 query ExampleQuery {
2   id
3 }
4
```

Response

# Let's make this interactive

***All workshop materials  
can be found here:***



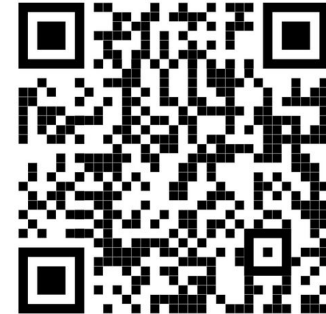
<https://shorturl.at/kvzIZ>  
<https://shorturl.at/bMU39>

***Some installation  
prerequisites for  
RaMP-DB:***



<https://shorturl.at/bgqrQ>

***Provide your  
comments/thoughts  
here:***



<https://shorturl.at/sS138>

## Filters

## Hierarchy

### Phenotypes

Search phenotypes

- ☐ Seizure 1006
- ☐ Intellectual disability 842
- ☐ Short stature 761
- ☐ Global developmental delay 723
- ☐ Microcephaly 579
- ☐ Scoliosis 565
- ☐ Hypotonia 525

### Genes

Search genes

Rare disease search



Sort

Articles

Items per page:

10

1 - 10 of 12004



### Glioblastoma: GARD:0002491

[Sign In](#)



[Disorder](#)

[Disease](#)

Synonyms: GBM, Glioblastoma multiforme

Published Articles

1932

Funded Projects

6878

Clinical Trials

1144

Associated Genes

0

Phenotypes

15

### Amyotrophic lateral sclerosis: GARD:0005786

[Sign In](#)



[Disorder](#)

[Disease](#)

Synonyms: ALS, Charcot disease, Lou Gehrig disease

Published Articles

1774

Funded Projects

6020

Clinical Trials

659

Associated Genes

37

Phenotypes

23

# Registering



Sign In with Google



Sign In with Facebook



Use an Email and Password

By continuing, you are indicating that you accept our [Terms of Service](#) and [Privacy Policy](#).



Enter your email\*

Password

[Forgot password?](#)

Sign In

Don't have an account?

[Register Now](#)

By continuing, you are indicating that you accept our [Terms of Service](#) and [Privacy Policy](#).

huntingt



Huntington disease

Juvenile Huntington disease

Huntington disease-like syndrome

Huntington disease-like syndrome due to C9ORF72 expansions

Spinocerebellar ataxia type 17

# Filter

Clear All 

## Phenotypes

Search phenotypes

- ☒ Intellectual disability 842
- ☒ Short stature 761
- ☒ Scoliosis 565
- ☐ Seizure 1006
- ☐ Global developmental delay 723
- ☐ Microcephaly 579
- ☐ Hypotonia 525


## Genes

Search genes

- ☒ COL2A1 20

## Selected Filters:

phenotypes  genes 

Intellectual disability 

COL2A1 

Short stature 

Scoliosis 

Sort

Articles 

Items per page:

10 

1 - 8 of 8



## Legg-Calvé-Perthes disease: GARD:0006874

Sign In



Disorder

Disease

Synonyms: Aseptic necrosis of the capital femoral epiphysis, Osteochondrosis of the capital femoral epiphysis, Perthes disease

Published Articles

690

Funded Projects

0

Clinical Trials

15

Associated Genes

1

Phenotypes


8

## Spondyloepiphyseal dysplasia congenita: GARD:0004987

Sign In





 Rare Disease Alert System

DISEASES ABOUT APIs

Sign In

Filters

Hierarchy

> Rare neurologic disease

> Rare genetic disease

> Rare developmental defect during embryogenesis

> Rare skin disease

▼ Rare endocrine disease

> Rare disorder with hypergonadotropic hypogonadism

> Rare endocrine growth disease

▼ Rare adrenal disease

> Adrenal/paraganglial tumor

> Adrenogenital syndrome

▼ Cushing syndrome

> Rare disease with Cushing syndrome as a major feature

> ACTH-dependent Cushing syndrome

> ACTH-independent Cushing syndrome

> Cushing syndrome due to macronodular adrenal hyperplasia

Rare disease search

Clear All

Selected Filters:

parentId X

GARD:0006224 X

Sort Articles

Items per page: 10 1 - 10 of 24

Cushing syndrome: GARD:0006224

Sign In ?

Group of disorders Clinical group

Synonyms: Hyperadrenocorticism, Hypercortisolism

Published Articles

362

Funded Projects

357

Clinical Trials

83

Associated Genes

0

Phenotypes

0

Cushing syndrome due to macronodular adrenal hyperplasia: GARD:0010824

Sign In ?

Disorder Disease

Synonyms: Primary bilateral macronodular adrenal hyperplasia

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15

## Huntington disease: GARD:0006677

Huntington chorea

Sign In

?

### Associated Genes (2)

Download

1 - 2 of 2

< >

Gene	Gene Name	Association Type	Reference
<a href="#">HTT</a>	<a href="#">huntingtin</a>	Disease-causing germline mutation(s) (gain of function) in	<a href="#">PMID:20301482</a> <a href="#">PMID:21566141</a>
<a href="#">SLC2A3</a>	<a href="#">solute carrier family 2 member 3</a>	Modifying germline mutation in	<a href="#">PMID:24452335</a>

### Phenotypes (53)

Download

1 - 5 of 53

< >

Phenotype	Frequency	Evidence <sup>i</sup>	Reference
<a href="#">Chorea</a>	Very frequent (99-80%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>
<a href="#">Hyperreflexia</a>	Very frequent (99-80%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>
<a href="#">Mental deterioration</a>	Very frequent (99-80%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>
<a href="#">Staring gaze</a>	Frequent (79-30%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>
<a href="#">Speech articulation difficulties</a>	Frequent (79-30%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>
<a href="#">Poor fine motor coordination</a>	Frequent (79-30%)	Traceable Author Statement	<a href="#">PMID:20301482</a> <a href="#">PMID:28221321</a>

Epidemiology Articles 15

Non-Epidemiology Articles 389

NIH-funded Projects 1505

Clinical Trials 224

1 - 10 of 15

< >

## Fractures within 2 years of an obstetric hospitalization: analysis of nationwide administrative data in Japan.

*Journal of bone and mineral metabolism* Vol 40

Fushimi K, Miyasaka N, Obayashi S, Moriwaki M, Terauchi M, Toba M

Quality Management Center, Tokyo Medical and Dental University, Yushima 1-5-45, Bunkyo, Tokyo, 113-8510, Japan.

PMID: [35690967](#)  
DOI: [10.1007/s00774-022-01336-4](#)

First Published: Jun 12, 2022  
Cited By: 0

### Epidemiologies: 1

**Epidemiology Type:**  
incidence  
**Epidemiology Rate:**  
4.5/10,000  
**Location:**  
Japan  
**Sex Affected:**  
women

Show Abstract

### Keywords

Administrative Database, Fracture, Osteoporosis, Lactation, Pregnancy

### Other Diseases Mentioned: 1

[Cushing syndrome](#)

### Annotations: 5

[Chemical](#)

steroid

[Disease](#)

### Mesh Terms: 10

Adult  
Bone Density  
Female  
Fractures, Bone  
Hospitalization

# Neo4j/Cypher

```
grant$ MATCH p=(n:GARD)-[r:RESEARCHED_BY]-(pr:Project) WHERE n.GardId IN ["GARD:0010510","GARD:0006677"] AND pr.funding_ye...
```



Graph



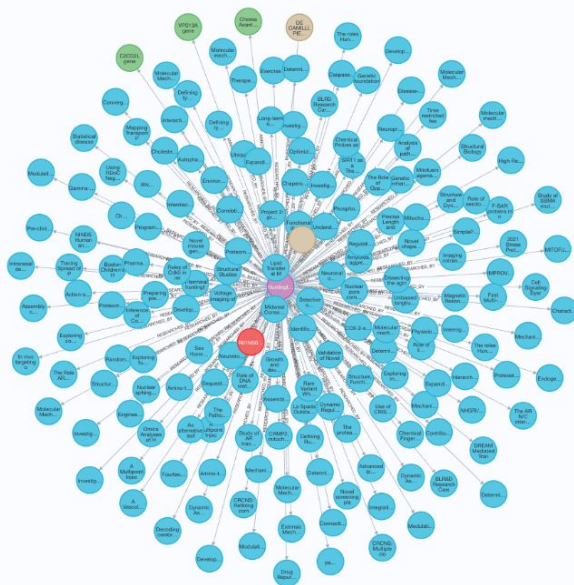
Table



Text



Code



Node properties

GARD

<id> 1412836  
GardId GARD:0006677  
GardName Huntington disease  
Synonyms [Huntington chorea]

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# Learn more about RDAS

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**Wednesday, Nov 15, 2023 9:45 AM - 11:00 AM CST**

## **PARENT SESSION**

Natural Language Processing - "Language City"

Location: Churchill B2



<https://shorturl.at/ftCFJ>



# Acknowledgements

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**NIH STRIDES Team**

**The Office of Data Science Strategy (ODSS) at NIH**

# Thank you!

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