Rare Disease Alert System (RDAS)

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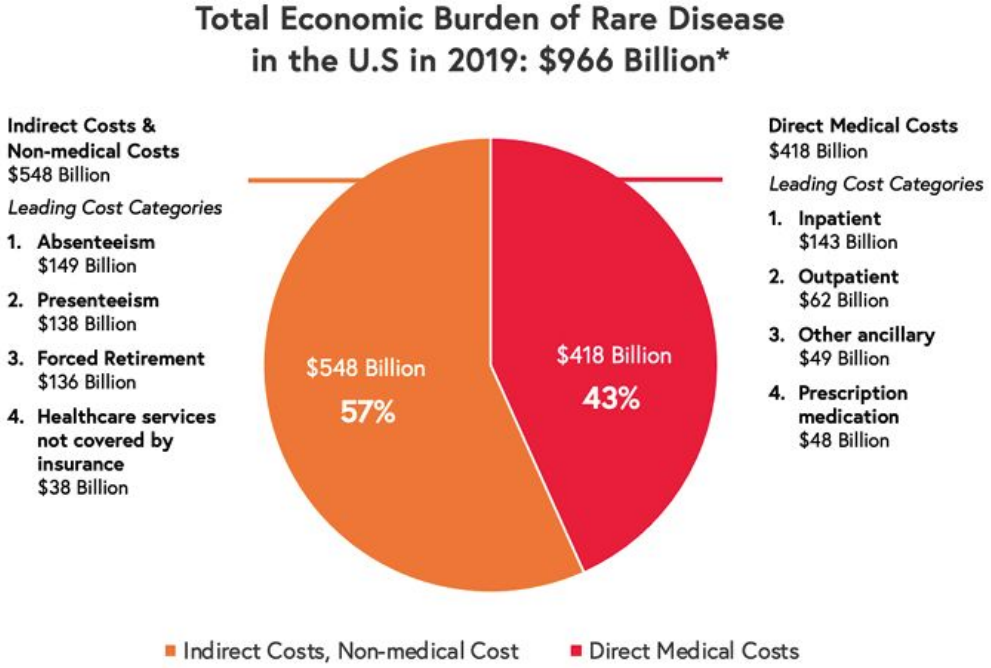
#AMIA2023

Burden of Rare Diseases



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

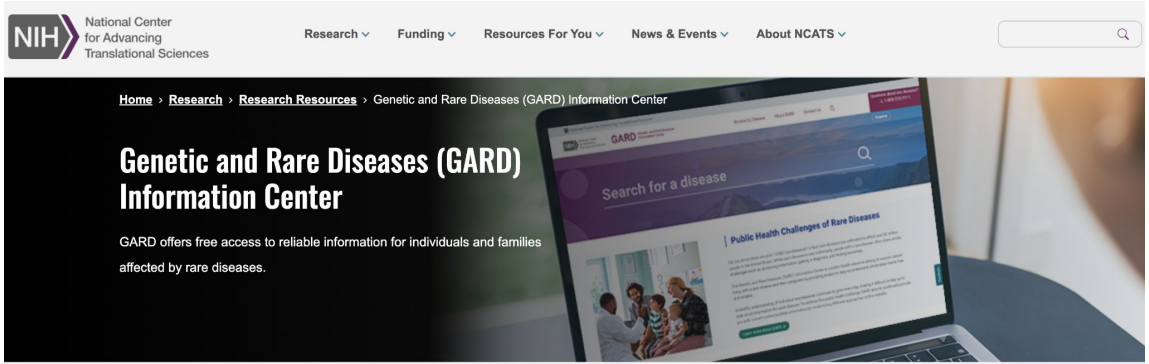
Genetic And Rare Diseases (GARD) Program

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

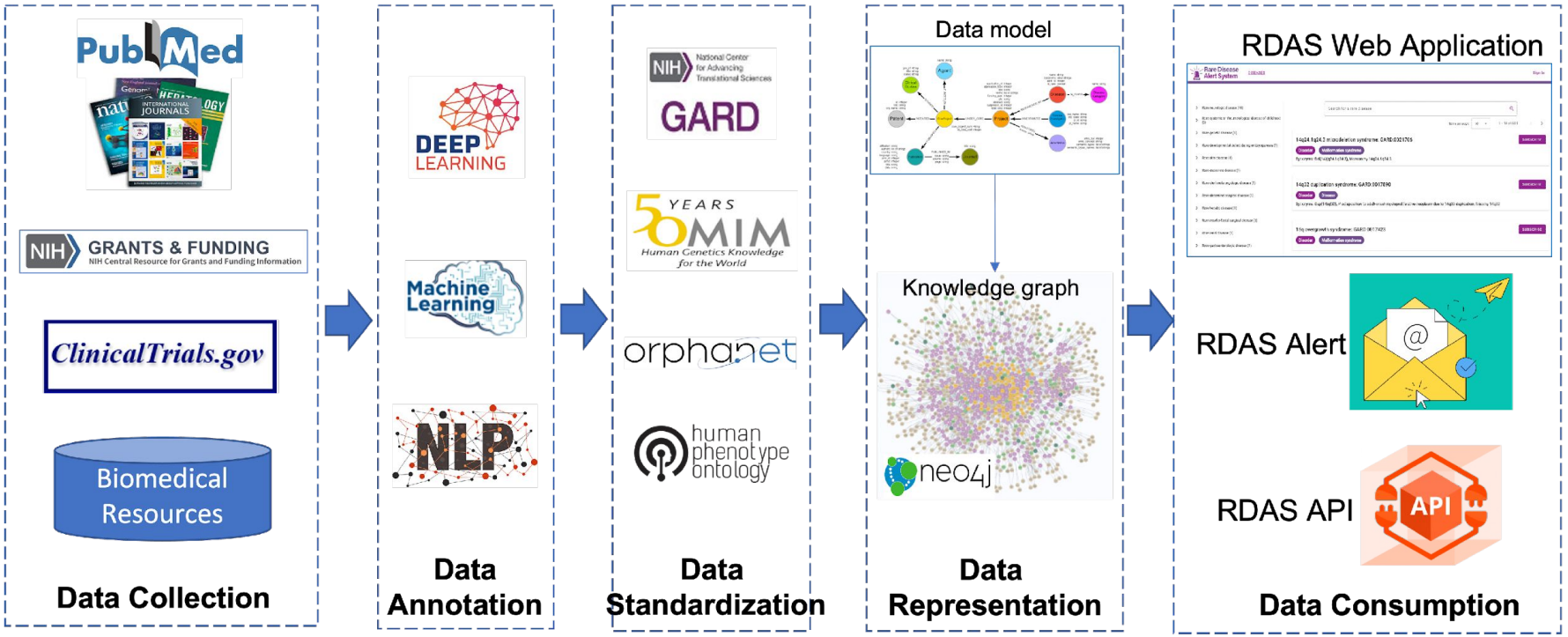
Our Goal: To empower the research community in their rare disease research efforts and to provide an educational resource

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

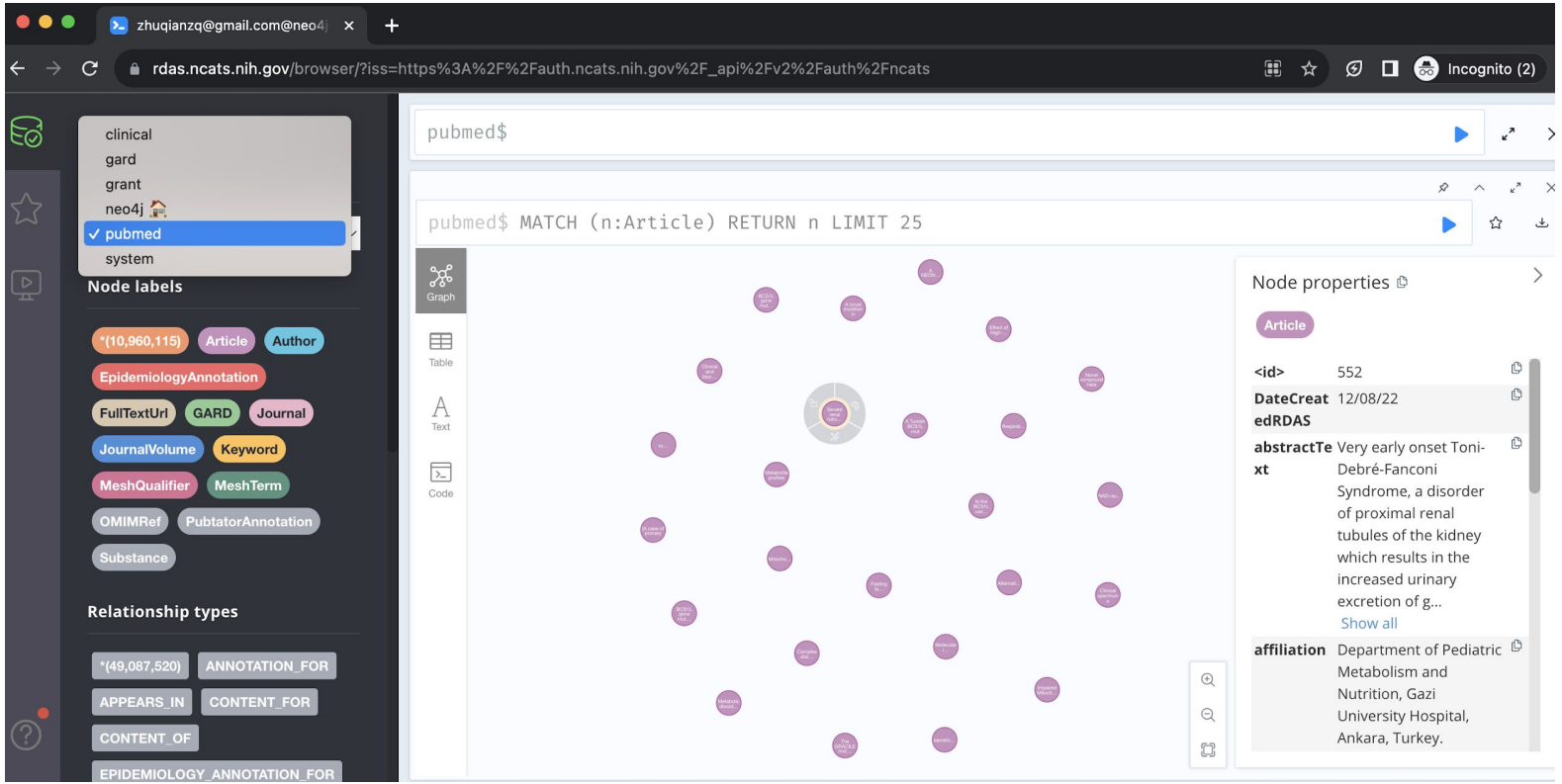
Main Features

- 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

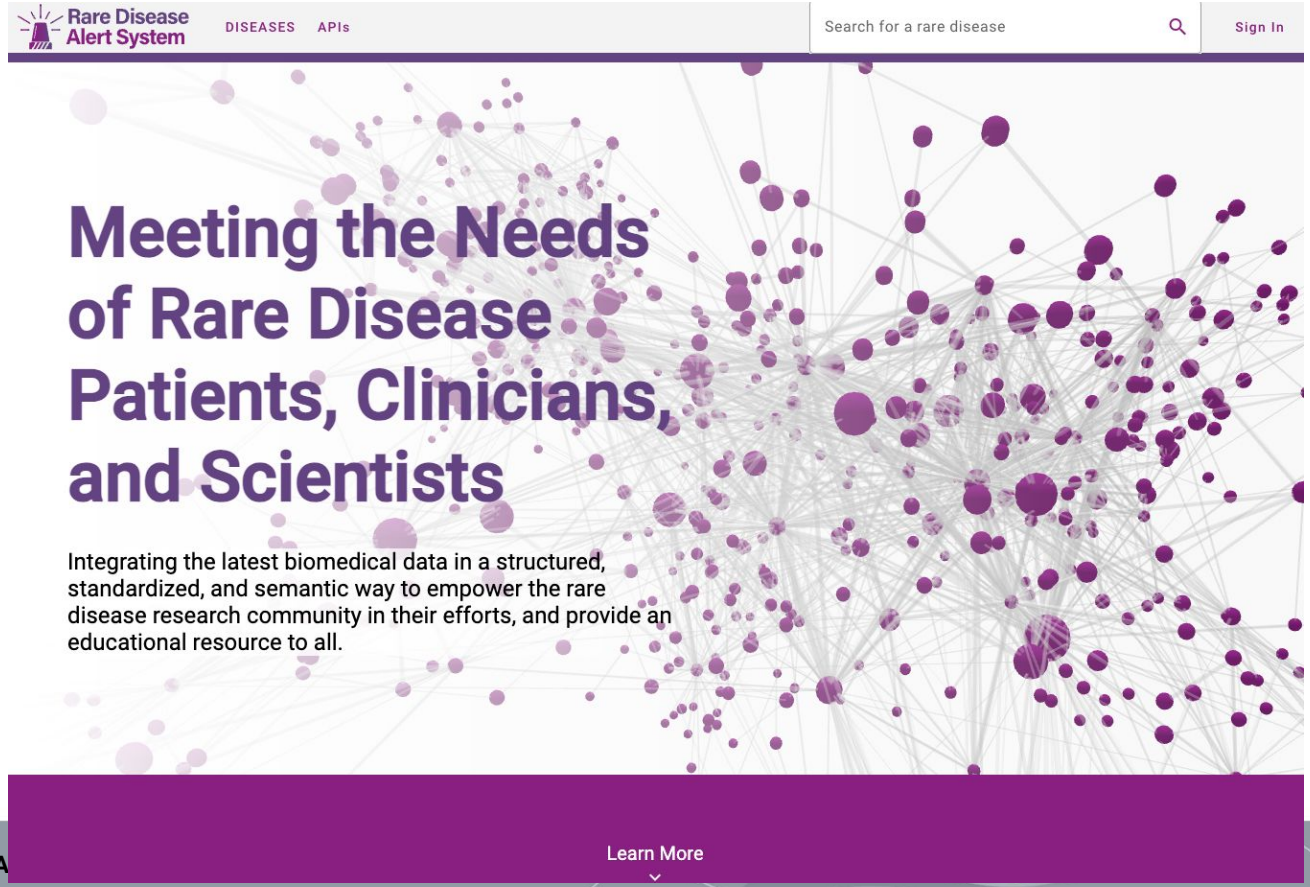
RDAS Architecture



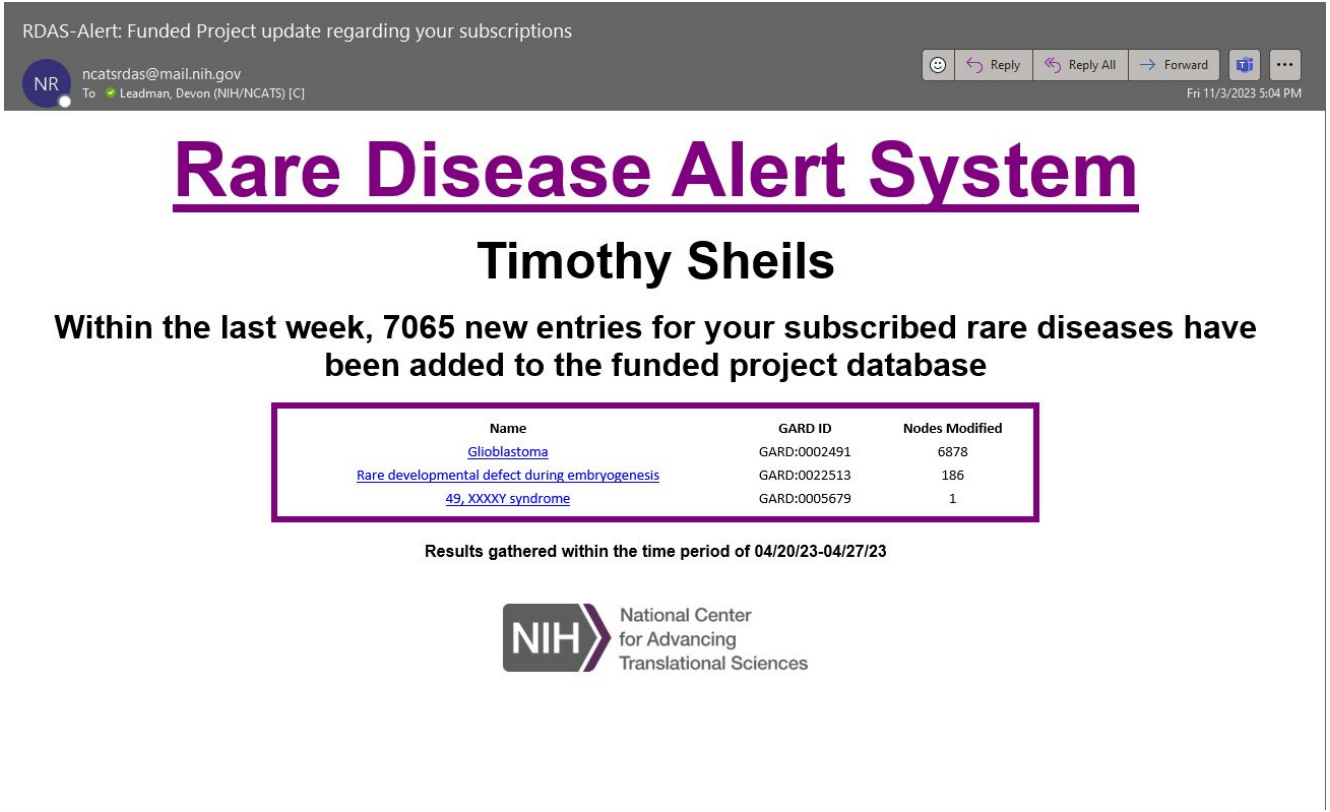
RDAS neo4j – https://rdas.ncats.nih.gov/browser



RDAS UI - https://rdas.ncats.nih.gov/



RDAS Alert



RDAS-Alert: Funded Project update regarding your subscriptions

NR 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
Glioblastoma	GARD:0002491	6878
Rare developmental defect during embryogenesis	GARD:0022513	186
49, XXXXY syndrome	GARD:0005679	1

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

NIH National Center for Advancing Translational Sciences

RDAS APIs

APIs

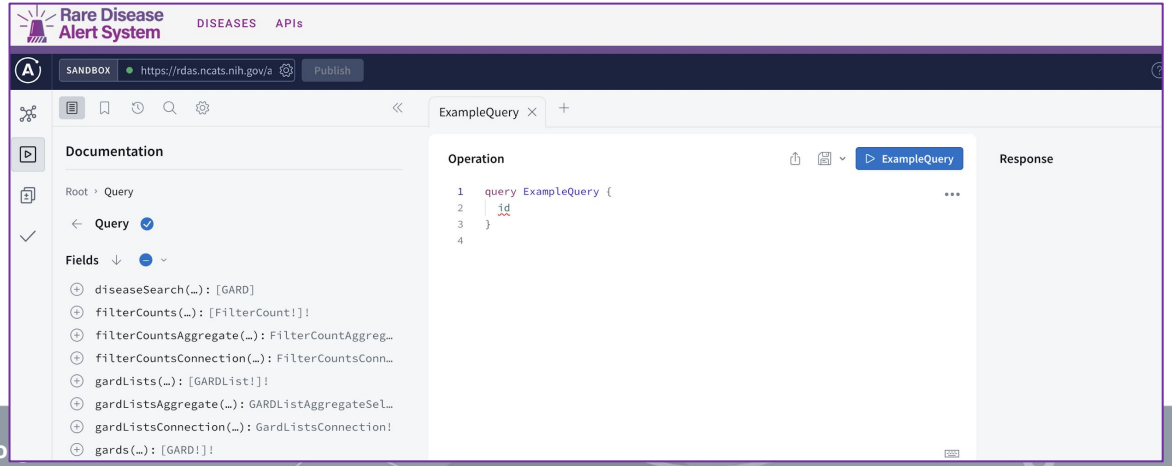
EPIDEMIOLOGY

DISEASES

PUBLICATIONS

PROJECTS

TRIALS



Let's make this interactive

All workshop materials can be found here:

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

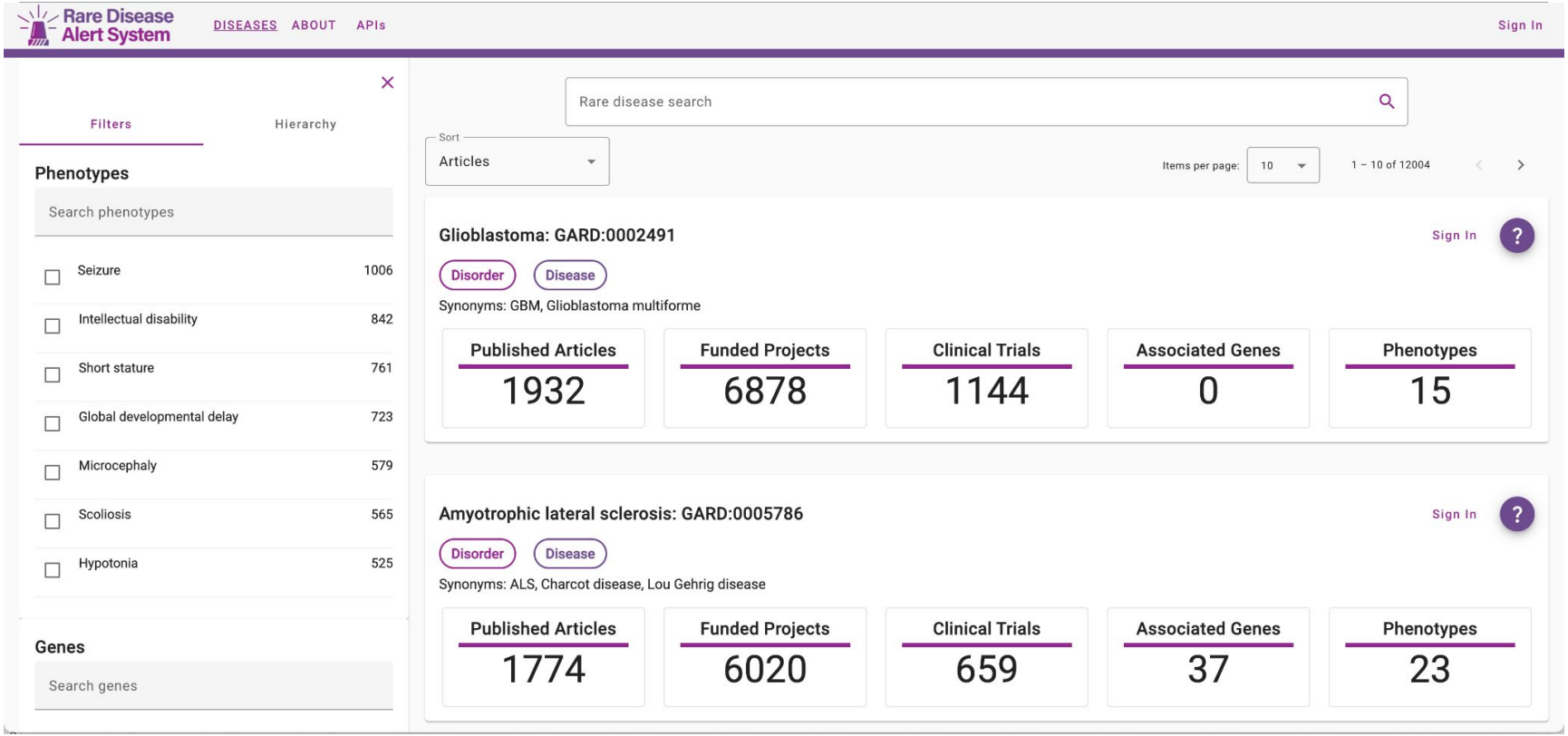
Some installation prerequisites for RaMP-DB:

https://shorturl.at/bgqrQ

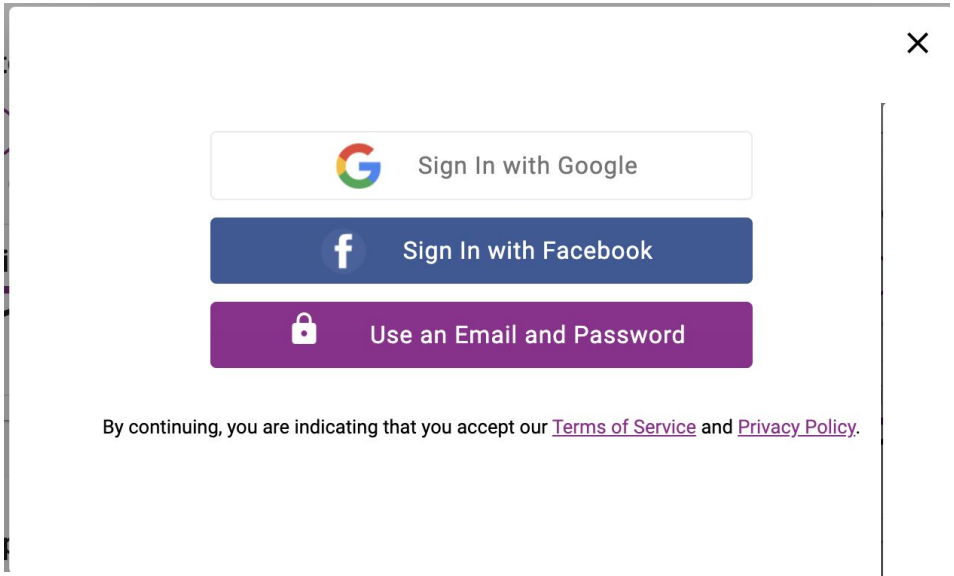
Provide your comments/thoughts here:

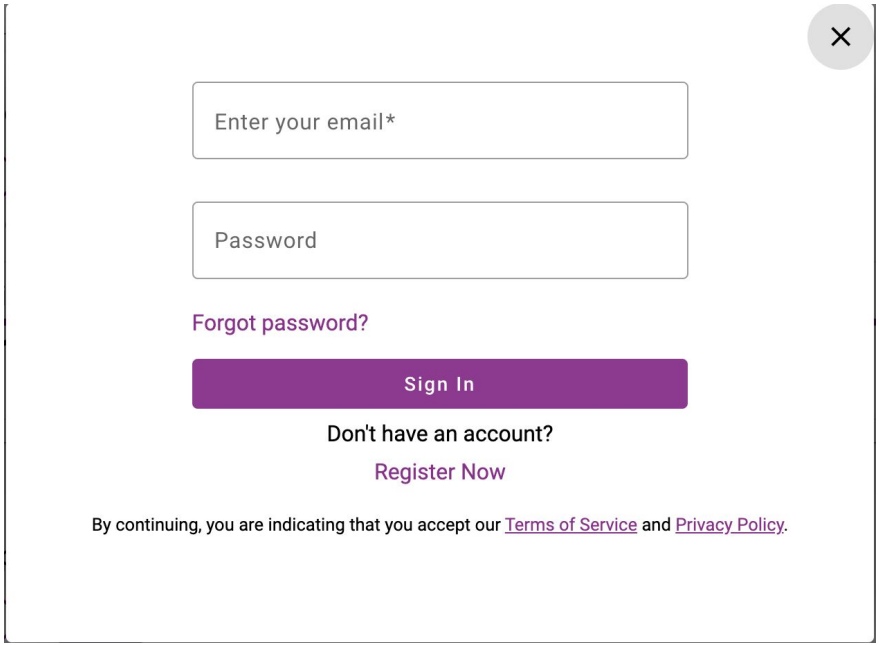
https://shorturl.at/sS138

Browse

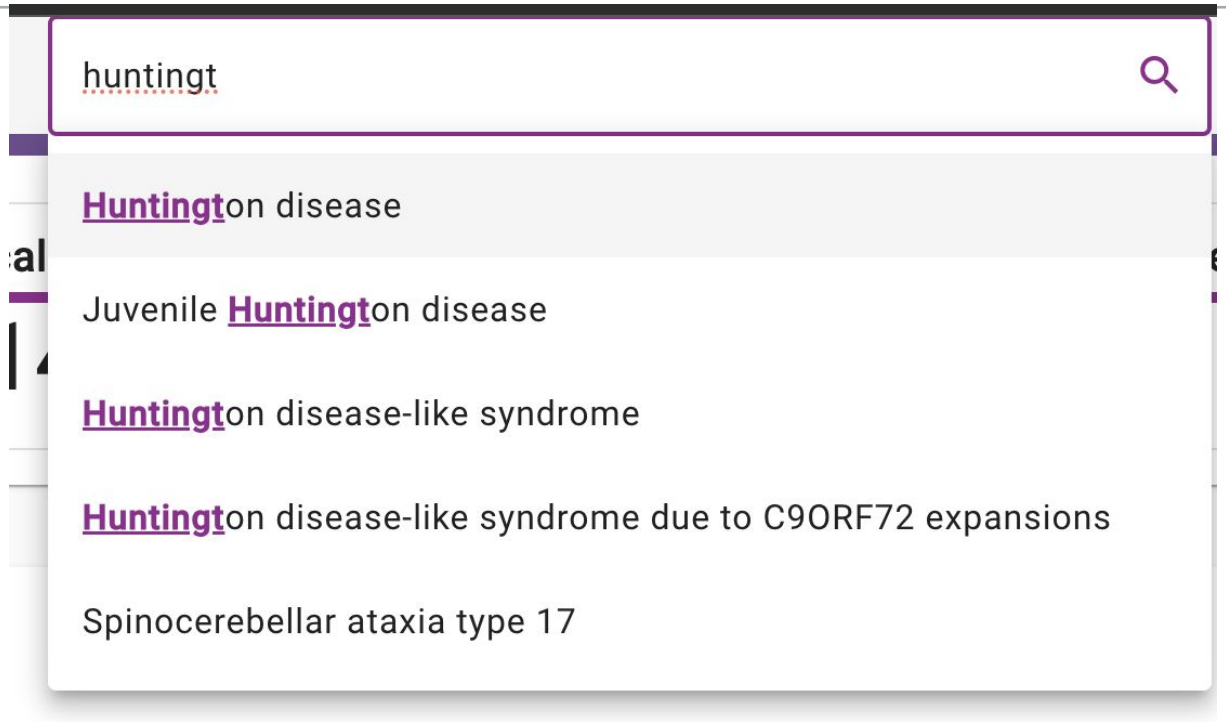

Rare Disease Alert System
DISEASES
ABOUT
APIs
Sign In
Filters
Hierarchy
Phenotypes
Search phenotypes
Seizure 1006
Intellectual disability 842
Short stature 761
Global developmental delay 723
Microcephaly 579
Scoliosis 565
Hypotonia 525
Genes
Search genes
Rare disease search
Sort
Articles
Items per page: 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

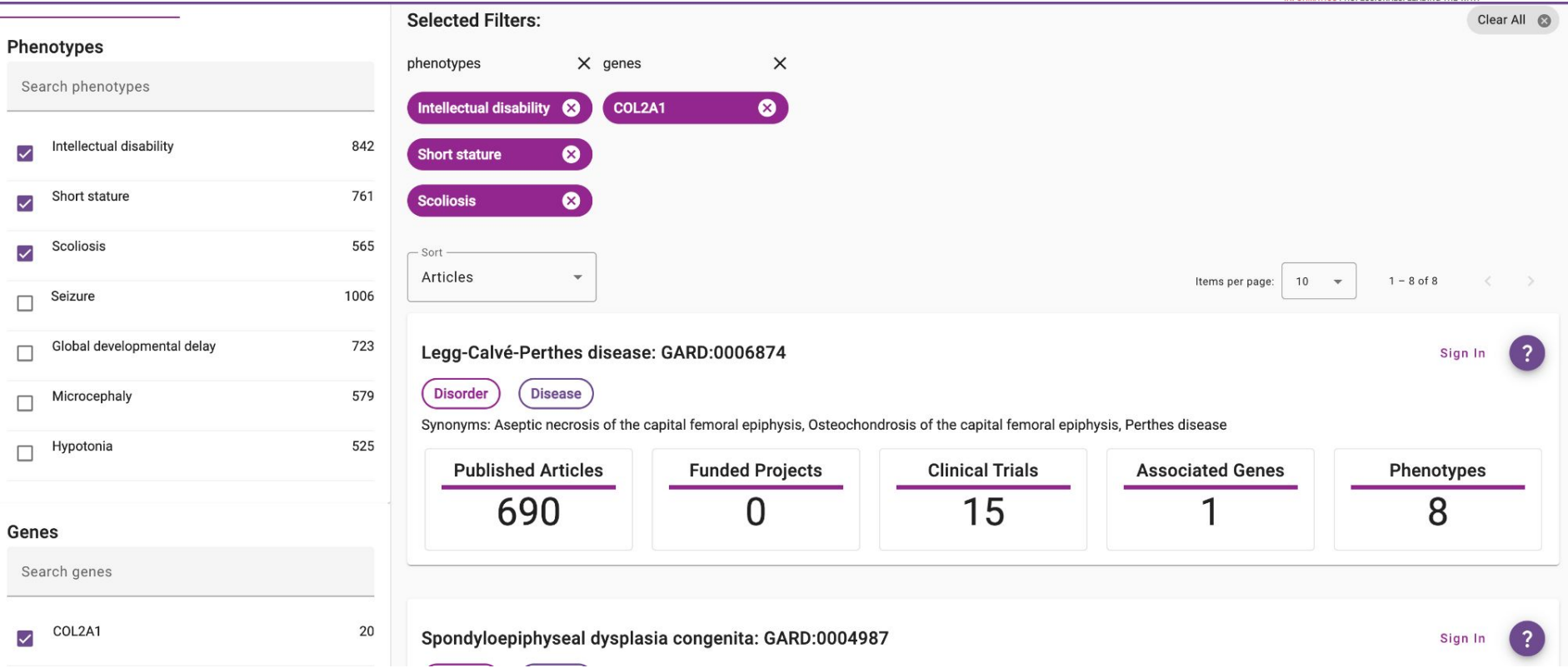




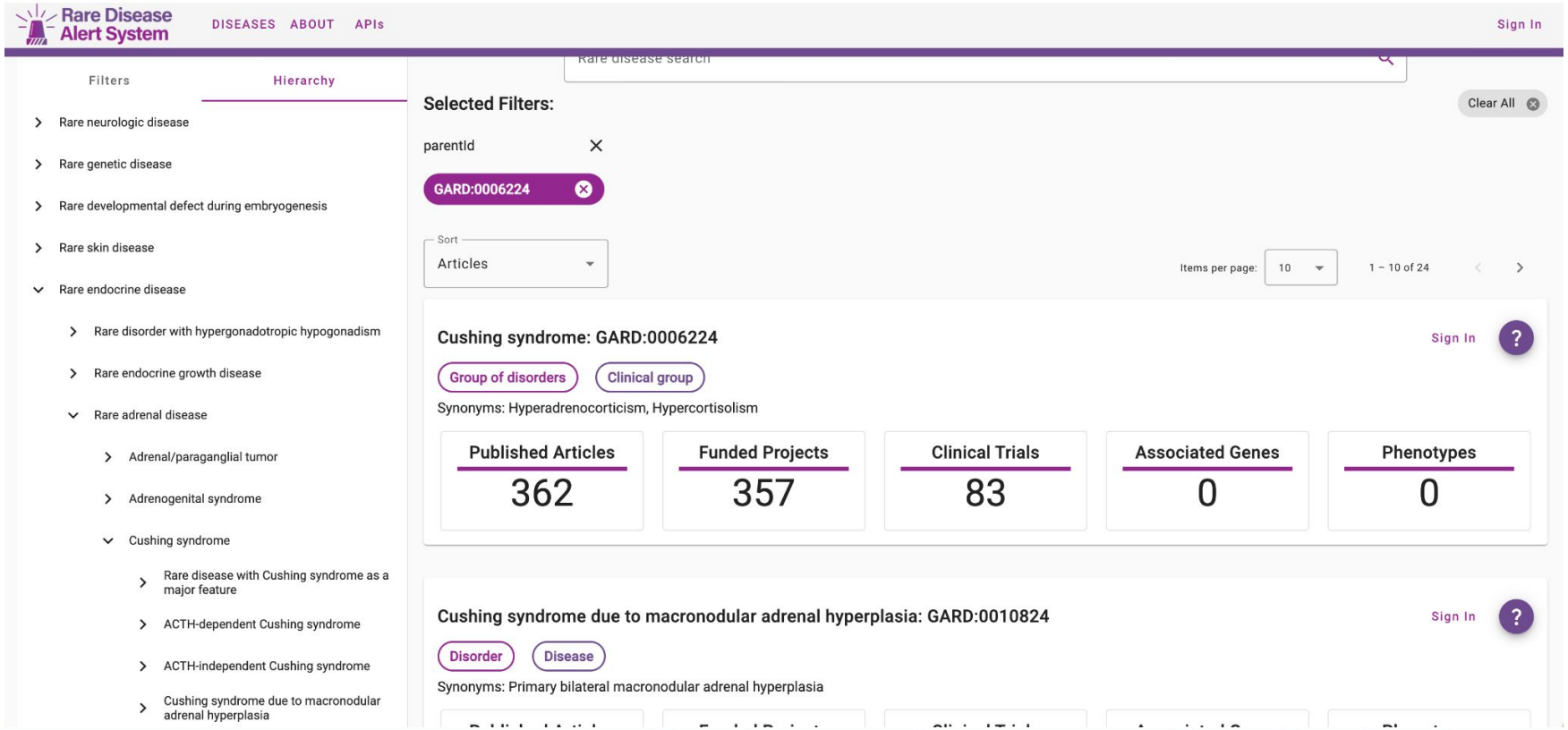
Search


huntingt
Huntington disease
Juvenile Huntington disease
Huntington disease-like syndrome
Huntington disease-like syndrome due to C9ORF72 expansions
Spinocerebellar ataxia type 17

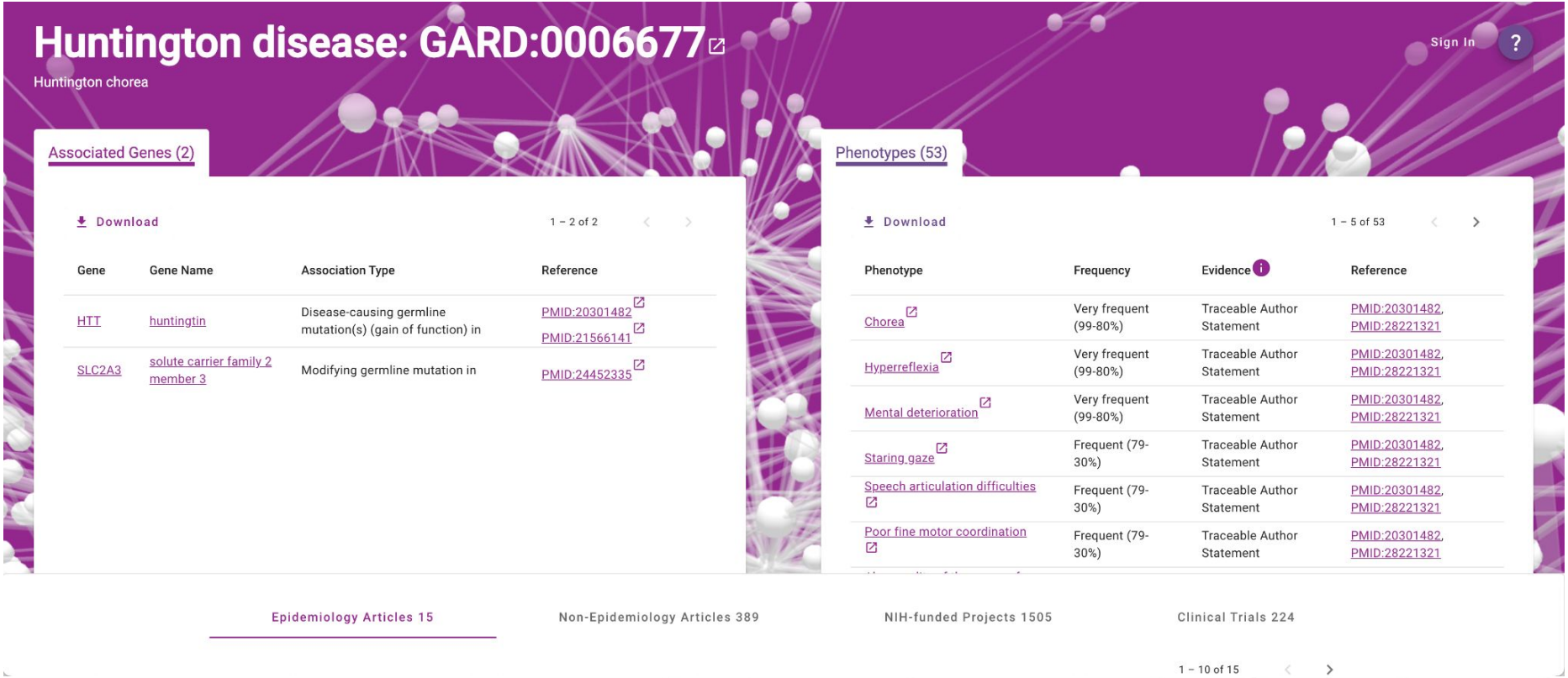
Filter


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:
Clear All
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

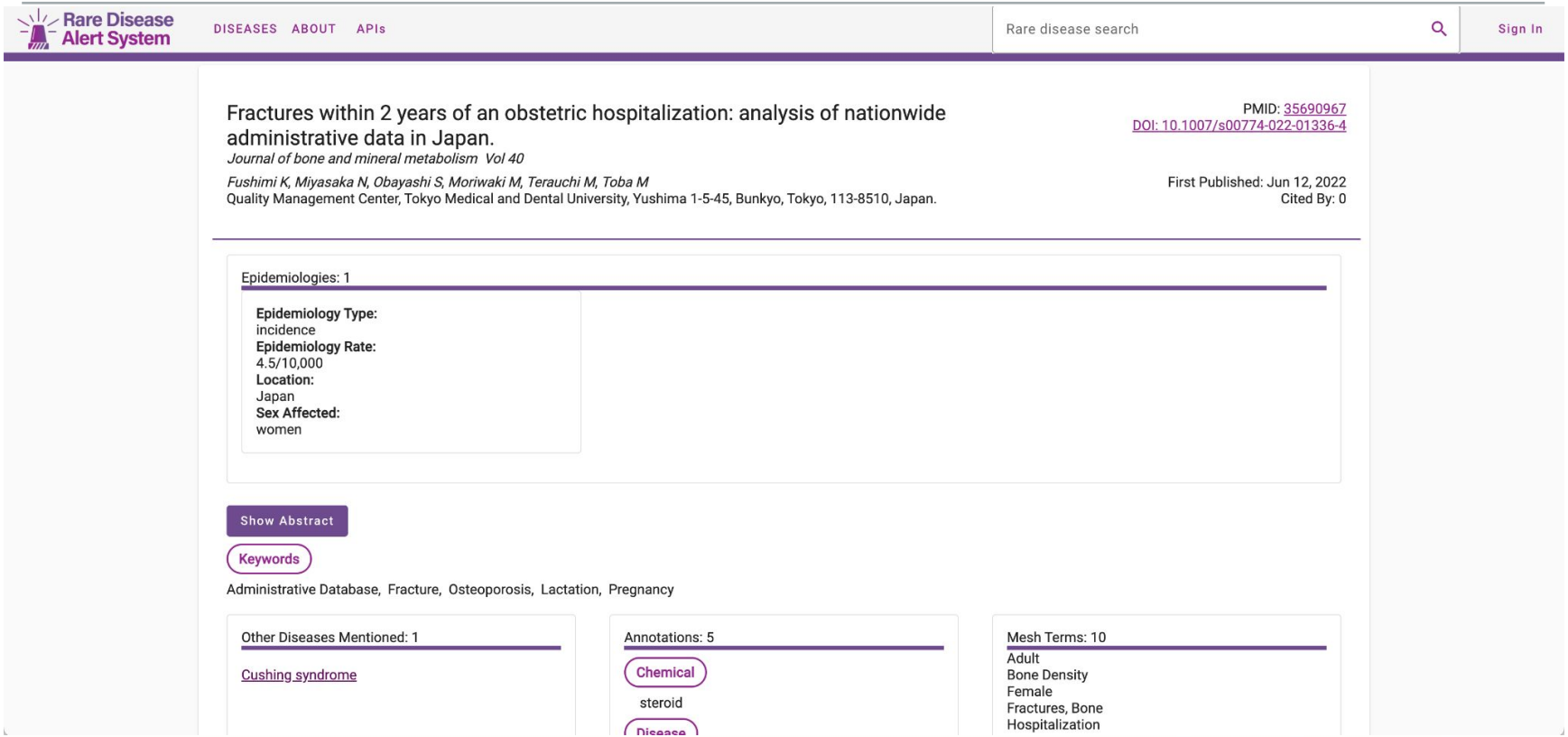
Hierarchy


Rare Disease Alert System
DISEASES
ABOUT
APIs
Sign In
Filters
Hierarchy
Rare disease search
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
Selected Filters:
Clear All
parentId
GARD:0006224
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

Details


Huntington disease: GARD:0006677
Huntington chorea
Sign In
Associated Genes (2)
Download
1 – 2 of 2
Gene
Gene Name
Association Type
Reference
HTT
huntingtin
Disease-causing germline mutation(s) (gain of function) in
PMID:20301482
PMID:21566141
SLC2A3
solute carrier family 2 member 3
Modifying germline mutation in
PMID:24452335
Phenotypes (53)
Download
1 – 5 of 53
Phenotype
Frequency
Evidence
Reference
Chorea
Very frequent (99-80%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Hyperreflexia
Very frequent (99-80%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Mental deterioration
Very frequent (99-80%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Staring gaze
Frequent (79-30%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Speech articulation difficulties
Frequent (79-30%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Poor fine motor coordination
Frequent (79-30%)
Traceable Author Statement
PMID:20301482, PMID:28221321
Epidemiology Articles 15
Non-Epidemiology Articles 389
NIH-funded Projects 1505
Clinical Trials 224
1 – 10 of 15

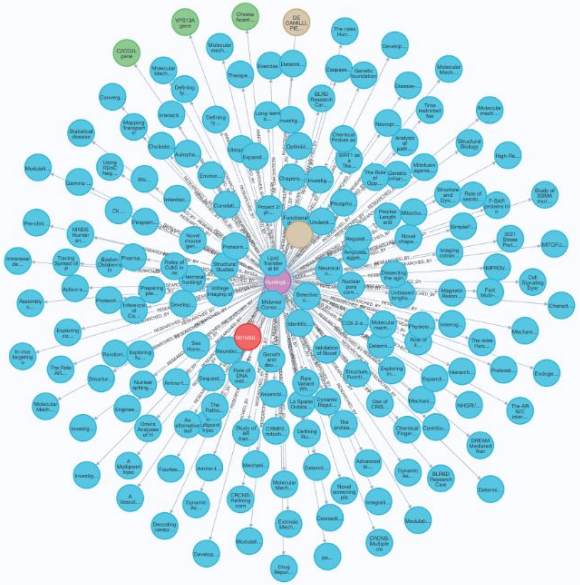
Epidemiology



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

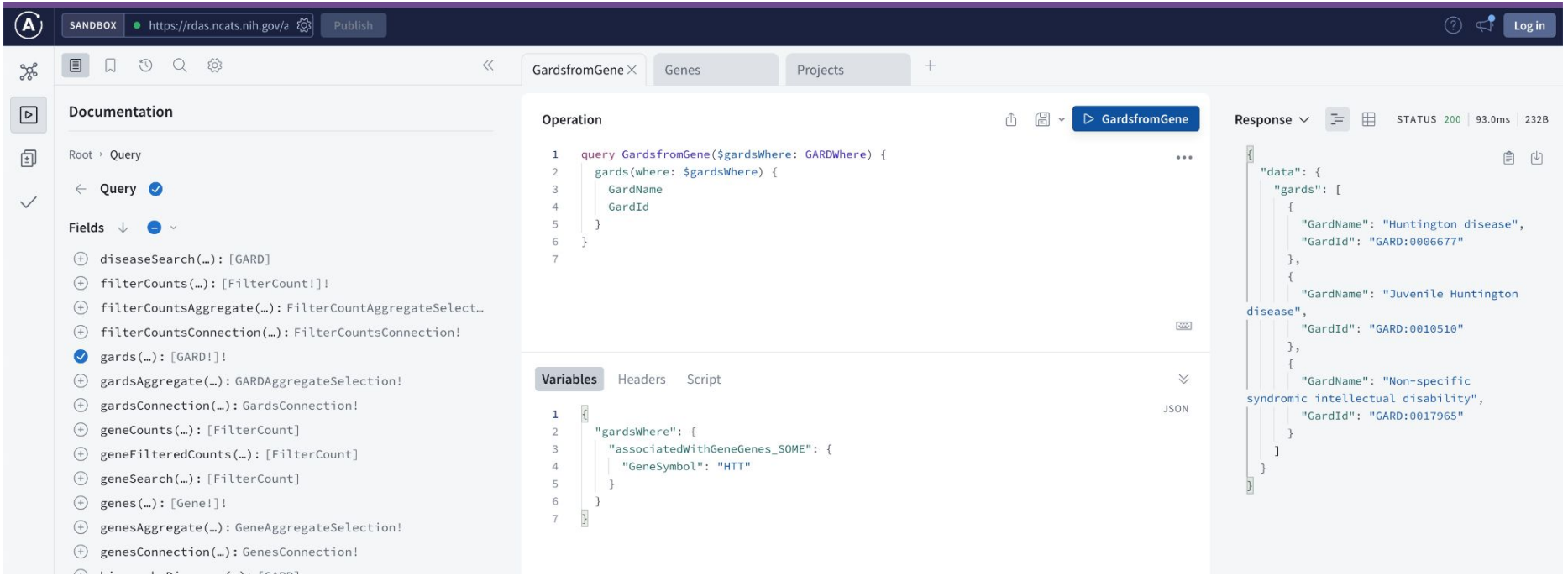


Node properties

GARD

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

GraphQL


SANDBOX
https://rdas.ncats.nih.gov/a
Publish
Log in
GardsfromGene
Genes
Projects
Documentation
Root › Query
Query
Fields
diseaseSearch(…): [GARD]
filterCounts(…): [FilterCount!]!
filterCountsAggregate(…): FilterCountAggregateSelect…
filterCountsConnection(…): FilterCountsConnection!
gards(…): [GARD!]!
gardsAggregate(…): GARDAggregateSelection!
gardsConnection(…): GardsConnection!
geneCounts(…): [FilterCount]
geneFilteredCounts(…): [FilterCount]
geneSearch(…): [FilterCount]
genes(…): [Gene!]!
genesAggregate(…): GeneAggregateSelection!
genesConnection(…): GenesConnection!
Operation
GardsfromGene
query GardsfromGene($gardsWhere: GARDWhere) {
gards(where: $gardsWhere) {
GardName
GardId
}
}
Variables
Headers
Script
JSON
{
"gardsWhere": {
"associatedWithGeneGenes_SOME": {
"GeneSymbol": "HTT"
}
}
}
Response
STATUS 200
93.0ms
232B
{
"data": {
"gards": [
{
"GardName": "Huntington disease",
"GardId": "GARD:0006677"
},
{
"GardName": "Juvenile Huntington disease",
"GardId": "GARD:0010510"
},
{
"GardName": "Non-specific syndromic intellectual disability",
"GardId": "GARD:0017965"
}
]
}
}

Learn more about RDAS

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

Informatics Core: Timothy Sheils, Shixue Sun, Ewy Mathé

Division of Rare Disease Research Innovations (DRDRI): Devon Leadman, Yanji Xu

Information Resources Technology Branch (ITRB): Carlin Biyoo, Eduaro Luiggi Lopez, Jennifer King, Hugo Hernandez, Sam Michael

NIH STRIDES Team

The Office of Data Science Strategy (ODSS) at NIH

Thank you!

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timothy.sheils@nih.gov