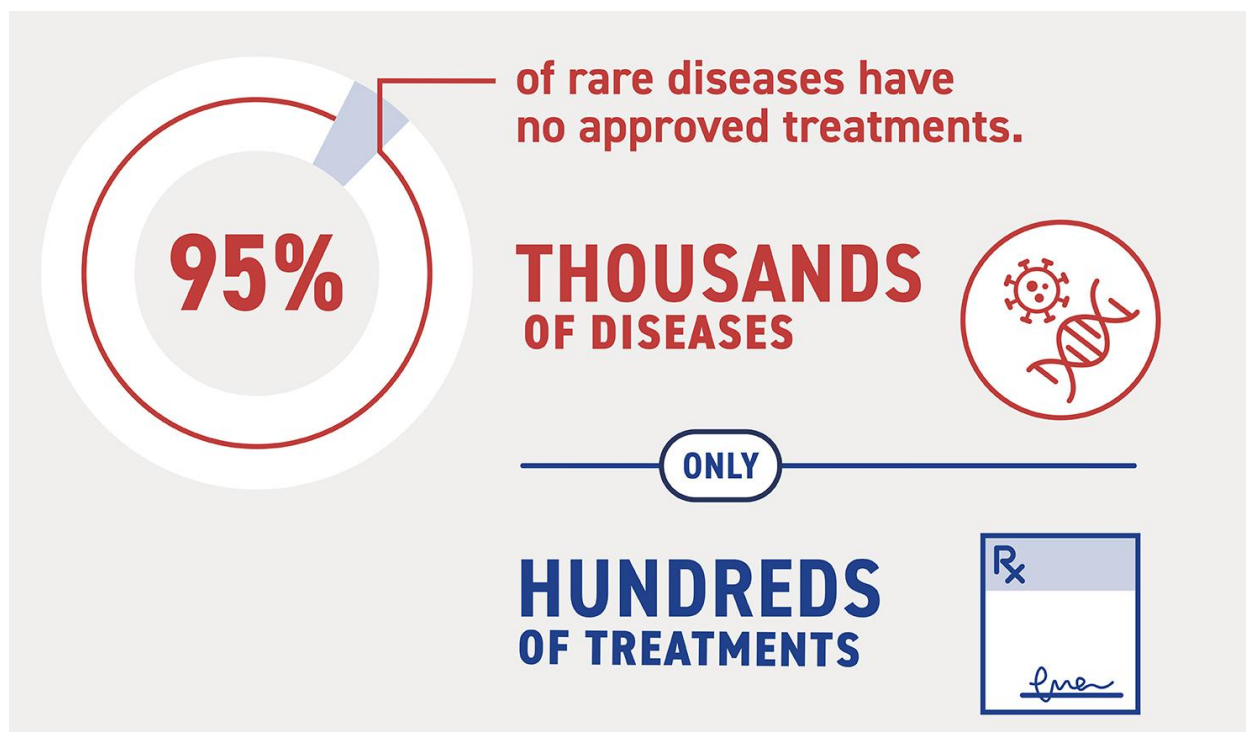


Transforming Data To Rare Disease Knowledge

Uncover insights into rare diseases to facilitate discoveries for new therapeutics



Approximately 80% of over 10,000 rare diseases have a genetic association but most lack approved treatments.

There are more than 10,000 known rare diseases and only a few hundred have safe, effective treatments. One challenge for researchers, patients, and other community partners is the ability to access and coordinate data from disparate sources where they are siloed, and not organized or harmonized, making it difficult to integrate in a useful manner.

The ability to make connections between rare diseases, associated gene variants, and phenotypes is critical for accelerating translational research and therapeutic development.

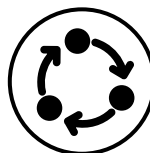
Leveraging the power of commonalities

To harmonize and integrate relevant siloed data sources, the Therapeutic Development Branch in the intramural Division of Preclinical Innovation at NCATS conceptualized RARe-SOURCE[®], an Integrated Bioinformatics Resource for Rare Diseases and in collaboration with the Advanced Biomedical and Computational Science developed and launched this platform for rare disease information.

The approach is to identify, extract, annotate, integrate, and enrich biomedical data from sources including the peer-reviewed scientific literature to help end-users uncover commonalities and share actionable knowledge that advances translational science and therapeutic discovery for rare diseases.



Integrate known public and private databases



Develop AI models for automating the mining of literature

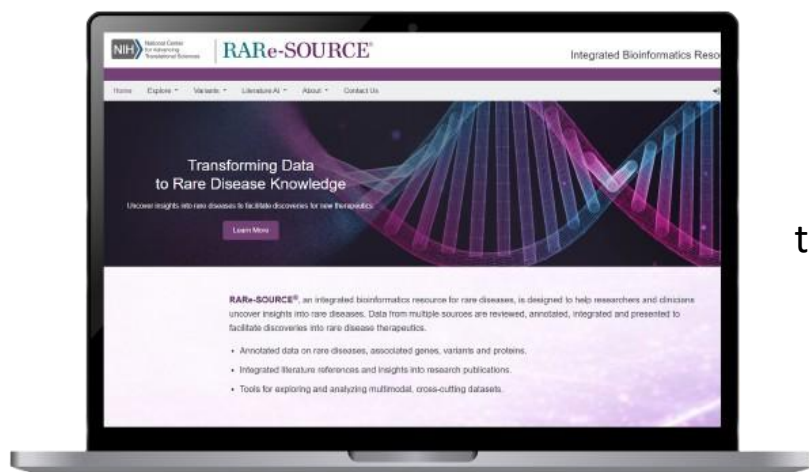


Use manual curation to understand gaps in knowledge



Expand to thousands of rare diseases

RARe-SOURCE®, an Integrated Bioinformatics Resource for Rare Diseases, provides an innovative, data-driven foundation to support scientific insight and translational impact.



Explore various tools through interactive tables and visualizations



Diseases

Search for information on rare diseases.



Curation

Examine manually curated variants and their clinical associations.



Genes

Explore genes associated with rare disease and related details.



Proteins

Visualize proteins related to rare diseases.



Variants

Review genetic variants and their impacts.



Literature

Access literature related to rare diseases.

Section Overview: Explore Disease and Gene

The Explore Diseases and Genes section was built on a carefully developed foundation. Disease and gene information collected from NCATS' Rare Diseases Informatics Platform (RDIP) team was integrated and harmonized with additional features developed by the team, including publication access, aliases search, external cross references, and data export capabilities. Together, these elements form a unified structure with information strategically connected to enable meaningful insights.

WHY THIS MATTERS

Designed with all users in mind, the interface makes it simple to search, filter, and export results without any technical background required. Regular updates provided by NCATS' RDIP team, combined with automated updates to additional disease details, ensure the information remains comprehensive and reliable, with latest research and expanding disease relationships are always reflected.

By the end of the section, you'll learn how to navigate the disease and gene tables and explore linked information and cross-referenced databases.



Find Disease Information

Search and filter through rare diseases to view detailed disease information.



Find Gene Information

Search and filter through genes to view detailed gene information.



Explore Connections

Efficiently connect rare disease details and relationships.

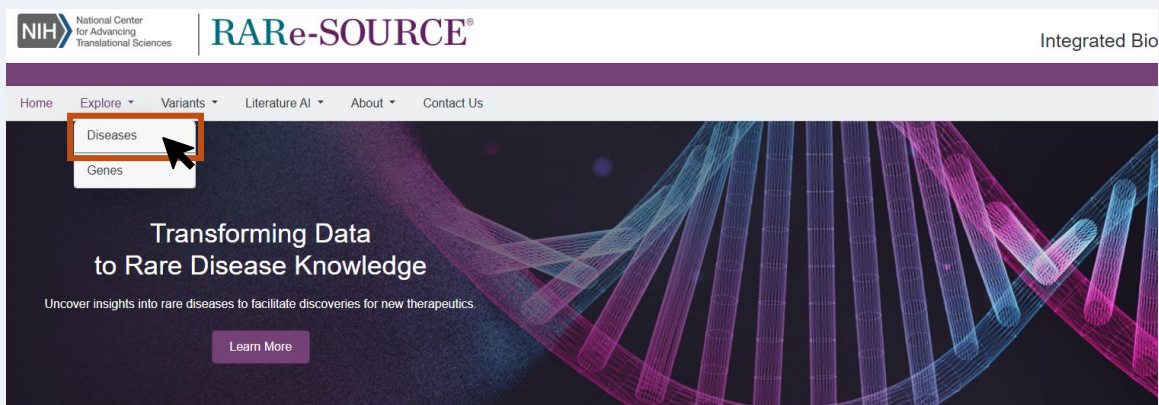


Export Findings

Download results to support your own research and analysis.

Example A: Exploring Diseases

- 1 Navigate to the “Explore” dropdown and select “Diseases” to launch the browse rare disease information page.



RARe-SOURCE®, an integrated bioinformatics resource for rare diseases, is designed to help researchers and clinicians uncover insights into rare diseases. Data from multiple sources are reviewed, annotated, integrated and presented to facilitate discoveries into rare disease therapeutics.

- Annotated data on rare diseases, associated genes, variants and proteins.
- Integrated literature references and insights into research publications.
- Tools for exploring and analyzing multimodal, cross-cutting datasets.

- 2 In the “Search” box, type in disease name. The results table shows disease information along with its synonyms, the associated gene(s), Literature AI links, cross-reference IDs, and additional links to other resources.

Browse Rare Disease Information

The RARe-SOURCE® platform integrates comprehensive information on rare diseases with known genetic associations. View detailed disease information and explore detailed disease information, associated genes, linked publications, and cross-references.

Search: Farber Disease Export Table Data 25 entries

Rare Disease Name	Associated Genes	Gene Disease Annotations	Disease Annotations	Disease IDs	Links
<input type="checkbox"/> Farber Lipogranulomatosis <input type="checkbox"/> Ac Deficiency <input type="checkbox"/> Acid Ceramidase Deficiency	ASAHI	■ Gene-Disease Literature AI	■ Disease Literature AI (234)	GARD: 0006426 OMIM: 228000 Orphanet: 333	■ PubMed

Showing 1 to 1 of 1 entry (filtered from 7,200 total entries)

Example A: Exploring Diseases (Cont'd)

- In the results table, select the disease name to open its information card in a pop-up window.

The screenshot shows the RARe-SOURCE search results page. At the top, there is a search bar containing 'Farber Disease' and an 'Export Table Data' button. Below the search bar is a table with columns: 'Rare Disease Name', 'Associated Genes', 'Gene Disease Annotations', 'Disease Annotations', and 'Dis'. The first row is highlighted and contains the following information:

Rare Disease Name	Associated Genes	Gene Disease Annotations	Disease Annotations	Dis
<input type="checkbox"/> Farber Lipogranulomatosis <ul style="list-style-type: none"> • Ac Deficiency • Acid Ceramidase Deficiency 	ASAH1	■ Gene-Disease Literature AI	■ Disease Literature AI (234)	GA OM Orp

Below the table, it says 'Showing 1 to 1 of 1 entry (filtered from 7,200 total entries)'. A red box highlights the 'Farber Lipogranulomatosis' text, and a black arrow points to it.

- The information card will display a list of known aliases, most recent publications tab, and its IDs with links to other related resources. To view in detail, see next page.

The screenshot shows the 'Rare Disease Information: Farber Lipogranulomatosis' page. It has two tabs: 'Disease Information' (selected) and 'Recent Publications'. The 'Disease Information' tab is active and displays the following sections:

- Known Disease Aliases:** A search bar and 'Export Table Data' button are at the top. Below is a list of 11 aliases: Ac Deficiency, Acid Ceramidase Deficiency, Acylsphingosine Deacylase Deficiency, Ceramidase Deficiency, Disseminated Lipogranulomatosis, Farber Disease, Farber's Disease, Farber's Lipogranulomatosis, Farber-uzman Syndrome, Fibril, and N-laurylsphingosine Deacylase Deficiency. It shows 'Showing 1 to 11 of 11 entries'.
- Disease ID Information:** A search bar and 'Export Table Data' button are at the top. Below is a table of IDs:

GARD	OMIM	Orphanet	UMLS	Mesh	ICD10CM
0006426	228000	333	C0268255	D055577	

It shows 'Showing 1 to 1 of 1 entry'.

Example A: Exploring Diseases (Cont'd)

Rare Disease Information: **Farber Lipogranulomatosis**

Disease Information Recent Publications

Known Disease Aliases Information provided by NIH National Center for Advancing Translational Sciences

Search: Export Table Data 25 entries per page

Disease Alias
Ac Deficiency
Acid Ceramidase Deficiency
Acylsphingosine Deacylase Deficiency
Ceramidase Deficiency
Disseminated Lipogranulomatosis
Farber Disease
Farber's Disease
Farber's Lipogranulomatosis
Farber-uzman Syndrome
Frbrl
N-laurylsphingosine Deacylase Deficiency

Showing 1 to 11 of 11 entries

Disease ID Information

Search: Export Table Data 25 entries per page

GARD	OMIM	Orphanet	UMLS	Mesh	ICD10CM
0006426	228000	333	C0268255	D055577	

1 Recent Publications
Access the three most recent publications related to the selected disease.

2 Aliases Search
Search through known aliases and synonyms to explore alternative names for the disease.

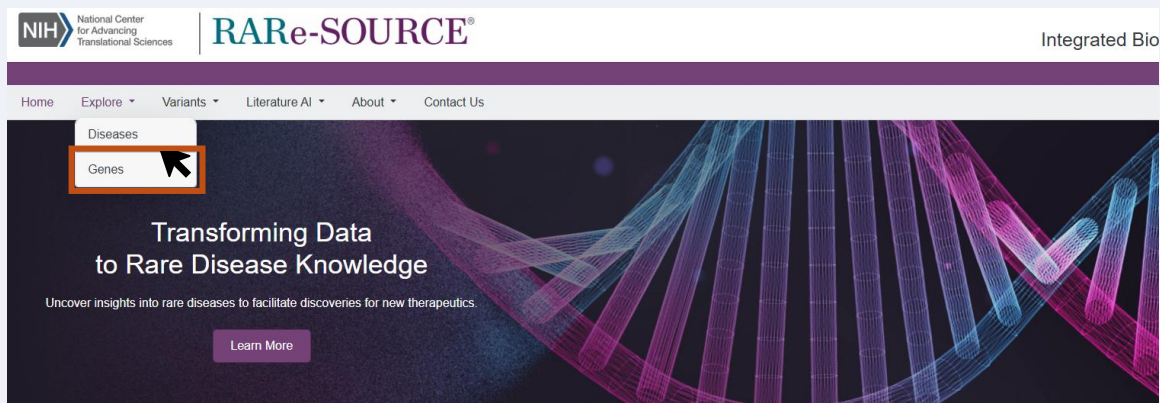
3 Export Disease Aliases
Download the full list of disease aliases as a CSV file.

4 Export Disease Identifiers
Download all linked resources IDs as a CSV file.

5 Disease Identifier Links Click any ID to be directed to its corresponding external resource.

Example B: Exploring Genes

- 1 Navigate to the “Explore” dropdown and select “Genes” to launch the browse gene information page.



RARe-SOURCE®, an integrated bioinformatics resource for rare diseases, is designed to help researchers and clinicians uncover insights into rare diseases. Data from multiple sources are reviewed, annotated, integrated and presented to facilitate discoveries into rare disease therapeutics.

- Annotated data on rare diseases, associated genes, variants and proteins.
- Integrated literature references and insights into research publications.
- Tools for exploring and analyzing multimodal, cross-cutting datasets.

- 2 In the “Search” box, type in gene name. The results table shows gene information along with its synonyms, the associated disease(s), Literature AI links, cross-reference IDs, and additional links to other resources.

Browse Genes Associated with Rare Diseases

The RARe-SOURCE® platform integrates comprehensive gene information associated with rare diseases. View and explore detailed gene information and explore associated variants, linked publications, and 2D/3D protein structures.

Search: Export Table Data 25 entries per

Gene Information	Associated Rare Diseases	Gene Disease Annotations	Gene Annotations	Gene IDs	Links
<input type="checkbox"/> SLC6A8 <input type="checkbox"/> Solute Carrier Family 6 Member 8	<ul style="list-style-type: none"> • Creatine Transporter Deficiency 	<ul style="list-style-type: none"> • Curated Variants (186) • Gene-Disease Literature AI 	<ul style="list-style-type: none"> • Variants (3871) • 3D Protein Structure • Gene Literature AI (1718) 	<ul style="list-style-type: none"> Entrez: 6535 Ensembl: ENSG00000130821 HGNC: 11055 	<ul style="list-style-type: none"> • PubMed • GARD Information

Showing 1 to 1 of 1 entry (filtered from 4,512 total entries) « < 1 > »

Example B: Exploring Genes (Cont'd)

- In the results table, select the gene name to open its information card in a pop-up window.

The screenshot shows the RARe-SOURCE website interface. At the top, there are navigation menus for Home, Explore, Variants, Literature AI, About, and Contact Us. Below the navigation is a section titled "Browse Genes Associated with Rare Diseases" with a brief description of the platform. A search bar contains the text "ASAH1" and an "Export Table Data" button is located to its right. The search results are displayed in a table with four main columns: "Gene Information", "Associated Rare Diseases", "Gene Disease Annotations", and "Gene Annotations". The first row of results is for the gene "ASAH1", which is highlighted with a red box and a black arrow. The "Gene Information" column shows "ASAH1" and "N-Acylsphingosine Amidohydrolase 1". The "Associated Rare Diseases" column lists "Spinal Muscular Atrophy-Progressive Myoclonic Epilepsy Syndrome", "Farber Lipogranulomatosis", and "ASAHI-Related Sphingolipidosis". The "Gene Disease Annotations" column shows "Gene-Disease Literature AI". The "Gene Annotations" column shows "Variants (15785)", "3D Protein Structure", and "Gene Literature AI (3950)".

- The information card will display the gene description, most recent publications tab, and associated diseases tab with IDs and links to other related resources. To view in detail, see next page.

The screenshot shows a detailed information card for the gene ASAH1. The card is titled "Gene Symbol: ASAH1" and contains the following information:

- Description:** N-Acylsphingosine Amidohydrolase 1
- Chromosomal Information:** chr8:18055992-18084998 (-), p22
- Related Diseases:** A tabbed interface with "Related Diseases" and "Recent Publications" tabs. The "Related Diseases" tab is active, showing a table of diseases associated with ASAH1. The table has columns for "Disease Name", "Disease ID", and "Class". The diseases listed are:

Disease Name	Disease ID	Class
Atrial Fibrillation	ME SH: D001281	
Farber Lipogranulomatosis	ME SH: D055077	
Lipidoses	ME SH: D008064	
Liver Cirrhosis, Experimental	ME SH: D008106	
Pneumocystosis	ME SH: D011009	
SPINAL MUSCULAR ATROPHY WITH PROGRESSIVE MYOCLONIC EPILEPSY	CMIM: 159950	
- GAD Disease Info:** A section for Gene Annotations with a search bar and an "Export Table Data" button. It shows a table of diseases associated with ASAH1:

Disease Name	Class	Disease ID
Tobacco Use Disorder	chemdependency	705318
Type 2 Diabetes edema rosiglitazone	pharmacogenomic	706217

Example B: Exploring Genes (Cont'd)

1 Description
N-Acylsphingosine Amidohydrolase 1

Chromosomal Information
chr8:18055992-18084998 (-); p22

Related Diseases

Recent Publications **2**

Comparative Toxicogenomics Database Information provided by **bioDBnet**
biological DataBase network

3 Search: **4** [Export Table Data](#) 25 entries per page

Disease Name	Disease ID
Atrial Fibrillation	MESH:D001281
Farber Lipogranulomatosis	MESH:D055577 5
Lipidoses	MESH:D008064
Liver Cirrhosis, Experimental	MESH:D008106
Pneumoconiosis	MESH:D011009
SPINAL MUSCULAR ATROPHY WITH PROGRESSIVE MYOCLONIC EPILEPSY	OMIM:159950

Showing 1 to 6 of 6 entries

GAD Disease Info

6 Search: [Export Table Data](#) 25 entries per page

Disease Name	Class	Disease ID
Tobacco Use Disorder	chemdependency	706318
Type 2 Diabetes edema rosiglitazone	pharmacogenomic	706317

1 Gene Description
View the full scientific name and description of the selected gene.

2 Recent Publications
Access top three most recent publications related to the selected gene.

3 CTD Search
Search related diseases linked to the gene provided by bioDBnet.

4 Export CTD
Download the full list of associated diseases and their IDs as a CSV file.

5 Disease ID Links
Click any ID to be directed to its corresponding external resource.

6 GAD Disease Search
Search disease associations and classifications sourced from GAD database.

Section Overview: Explore LiteratureAI

One approach to extracting data is developing Natural Language Processing models to automate the mining of scientific literature and other databases. The LiteratureAI module does exactly this – scanning titles and abstracts of papers from Medline, identifying mentions of rare diseases and their associated genes, and extracting articles for users to review.

WHY THIS MATTERS

Searching for specific rare disease literature can be overwhelming and time consuming. LiteratureAI is built to be accessible to anyone, automatically accounting for as many known disease aliases and gene synonyms to ensure relevant articles are not missed. Disease or Gene search options organize results by focusing on publications most relevant to a specific disease or gene, helping users identify key articles without manually sorting through large volumes of literature. Additionally, the combined Gene-Disease Co-occurrence feature adds another layer by connecting disease and gene mentions together, giving users a more concise and connected view of the literature in one place.

By the end of the section, you'll learn how to navigate the literature mining tool, discover relevant scientific publications on rare diseases and their associated genes.



Search Literature

Find publication by rare disease, gene, or disease-gene co-occurrence.



Identify Publication Trends

Use charts to track publication volume over time and across journals.



Refine Your Results

Sort and filter results easily without having to scroll through hundreds of results.

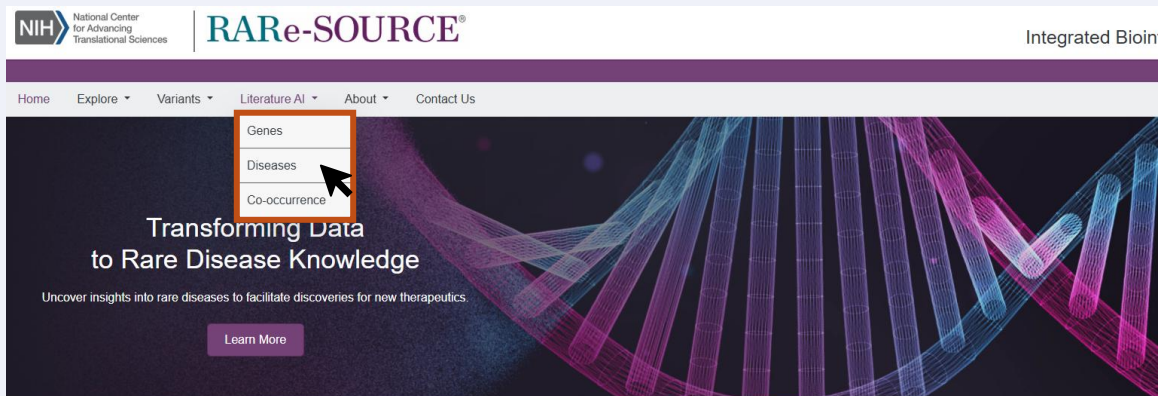


Export Findings

Download results to support your own research and analysis.

Example C: Explore Literature AI

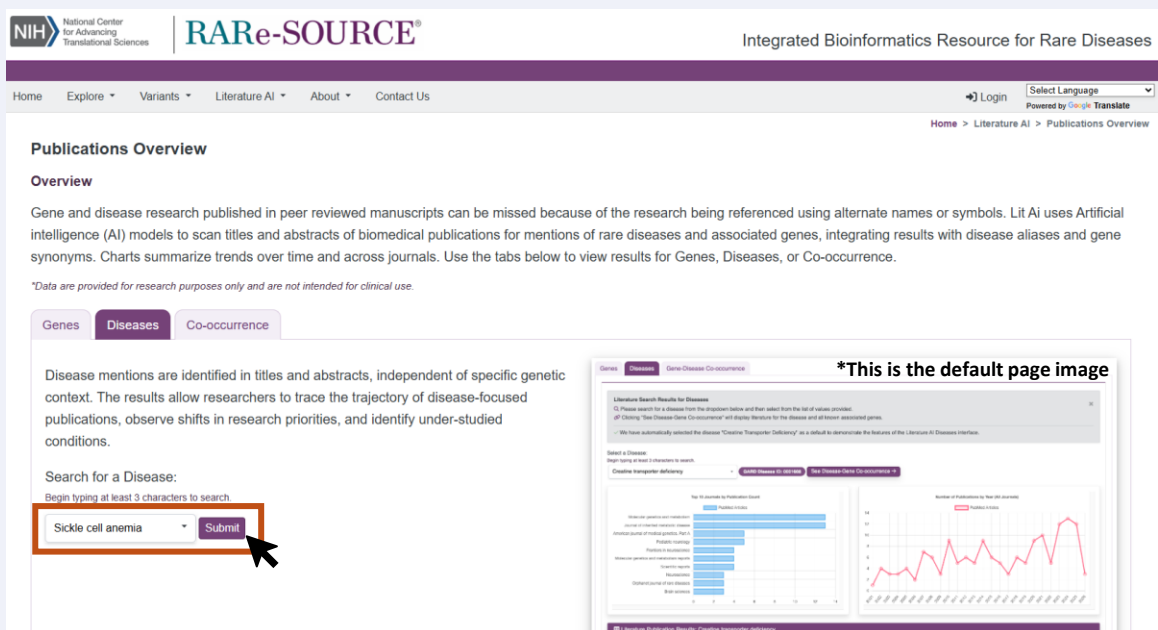
- 1 Navigate to the “LiteratureAI” dropdown and select “Diseases,” “Genes,” or “Co-occurrence” to launch the publications overview page.



RARe-SOURCE®, an integrated bioinformatics resource for rare diseases, is designed to help researchers and clinicians uncover insights into rare diseases. Data from multiple sources are reviewed, annotated, integrated and presented to facilitate discoveries into rare disease therapeutics.

- Annotated data on rare diseases, associated genes, variants and proteins.
- Integrated literature references and insights into research publications.
- Tools for exploring and analyzing multimodal, cross-cutting datasets.

- 2a For disease or gene searches, type the disease or gene name in the “Search Box.” Click the “Submit” button to view the results page.



Example C: Explore LiteratureAI (Cont'd)

2b For Co-occurrence searches (Disease/Gene), select the “Gene” or “Disease” button. Then, type in your selections in the fields below and click the "Submit" button to view the results page.

Publications Overview

Overview

Gene and disease research published in peer reviewed manuscripts can be missed because of the research being referenced using alternate names or symbols. Lit AI uses Artificial intelligence (AI) models to scan titles and abstracts of biomedical publications for mentions of rare diseases and associated genes, integrating results with disease aliases and gene synonyms. Charts summarize trends over time and across journals. Use the tabs below to view results for Genes, Diseases, or Co-occurrence.

Data are provided for research purposes only and are not intended for clinical use.

Genes Diseases **Co-occurrence**

Publications where both a rare disease and an associated gene are mentioned in the title or abstract. Because the AI search is looking for both disease and gene mentions, results here are more constrained; for broader discovery, start with the *Genes* or *Diseases* tabs.

Find co-occurring publications:

Start with: **Genes** **Diseases**

Disease
Begin typing at least 3 characters to search.
Sickle cell anemia

Gene
HBB

Submit **Clear**

The screenshot shows the search interface with the 'Co-occurrence' tab selected. The search criteria are 'Sickle cell anemia' for the disease and 'HBB' for the gene. The 'Submit' button is highlighted with a red box and an arrow. A secondary screenshot shows the search results page with two charts: 'Top 10 Journals by Publication Count' and 'Number of Publications by Year (All Journals)'. Both charts are also highlighted with red boxes and arrows.

3 At the top of the results page, you can optionally view the two charts to see publication trends of the top journals by article count and the number of publications by year.

Genes Diseases **Gene-Disease Co-occurrence**

Literature Search Results for Gene-Disease Co-occurrence

- Indicate whether your search will start with a gene, or with a disease.
- Type to search, or select from the list of genes or diseases provided.
- Select one or more values from the list of all known genes/diseases related to your selection in step 2, then click "Display".

1 Start with: **Genes** **Diseases** 2 Select Gene: **HBB** 3 Select Disease: **Hb SS disease** **Display** **Reset**

Currently displaying results for: **HBB and Hb SS disease**

Top 10 Journals by Publication Count

Journal	Publication Count
Hemoglobin	~45
Blood	~25
British journal of haematology	~15
The Cochrane database of systematic reviews	~10
PLoS one	~10
American journal of hematology	~10
National Academy of Sciences of the United States of America	~10
Blood cells, molecules & diseases	~10
Clinical chemistry	~10
International journal of molecular sciences	~10

Number of Publications by Year (All Journals)

The line chart shows a general upward trend in the number of publications over time, with a significant peak around 2020-2025. The y-axis represents the number of publications (0 to 30), and the x-axis represents the year (1971 to 2025).

Example C: Explore LiteratureAI (Cont'd)

- 4 Publications are shown in the results table. To filter the articles, scroll down to the "Search Results" box and type in a keyword.

Filters Active - 0 Collapse All Show All Clear All

Gene Term Types x Aa# v Disease Name Types x Aa# v Disease Names x Aa# v

Journal Name x Aa# v Publication Date x Aa# v

Search Results: Export Table Data 25 entries per page

Gene ID	Gene Term Types	Gene Terms	Disease ID	Disease Name Types	Disease Names	Article Title & Information
3043	GENE SYMBOL	HBB	0008614 GARD	Alias	Scd	New Born Screening of Hemoglobinopathies in a Center Tunisian Population. Journal: Journal of pediatric hematology/oncology (2024) PMID: 38748601
3043	GENE SYMBOL	HBB	0008614 GARD	Alias	<ul style="list-style-type: none"> Scd Sickle Cell Anemia Sickle Cell Disease 	The Alberta Newborn Screening Approach for Sickle Cell Disease: The Advantages of Molecular Testing. Journal: International journal of neonatal screening (2021) PMID: 34842602
3043	GENE SYMBOL	HBB	0008614 GARD	Alias	<ul style="list-style-type: none"> Scd Sickle Cell Disease 	Evaluation of Technical Issues in a Pilot Multicenter Newborn Screening Program for Sickle Cell Disease. Journal: International journal of neonatal screening (2019) PMID: 33072962

- 5 In the "Filter Table Data" section, you can refine results by gene or disease name, gene or disease type, journal name, or publication date.

Filter Table Data x

▼ Please use the filters below to search and filter the data presented in the table. Use the button controls near the filters to collapse/show, clear and sort filters alphabetically and numerically.
 ↔ Each filter applied will automatically update the values in the other filter panes and will update the table data immediately.
 ⚙️ You can select multiple values in a filter pane by holding the [Ctrl] key on Windows or the [command] key on Mac. You can select a range of values by holding the [Shift] key on your keyboard.

Filters Active - 4 Collapse All Show All Clear All

Gene Term Types x Aa# v Disease Name Types x Aa# v Disease Names x Aa# v

Journal Name x Aa# v Publication Date x Aa# v

2019 z
 2021 1
 2023 z
 2024 1
 2025 z

Search Results: Export Table Data 25 entries per page

Gene ID	Gene Term Types	Gene Terms	Disease ID	Disease Name Types	Disease Names	Article Title & Information
3043	GENE SYMBOL	HBB	0008614 GARD	Alias	<ul style="list-style-type: none"> Scd Sickle Cell Disease 	Newborn Screening for Sickle Cell Disease and Thalassemia. Journal: JAMA health forum (2025) PMID: 40953336
3043	GENE SYMBOL	HBB	0008614 GARD	Alias	Scd	New Born Screening of Hemoglobinopathies in a Center Tunisian Population. Journal: Journal of pediatric hematology/oncology (2024) PMID: 38748601
3043	GENE SYMBOL	HBB	0008614	Alias	<ul style="list-style-type: none"> Scd 	Scalable noninvasive amplicon-based precision sequencing (SNAPseq) for genetic diagnosis and screening of β-thalassemia and sickle cell disease

Example C: Explore LiteratureAI (Cont'd)

6 After using the search, sort, and filter features, click on the article title in the results table to access the publication in PubMed. Note that some articles are freely available, while others may require subscription access.

Filter Table Data
 Please use the filters below to search and filter the data presented in the table. Use the button controls near the filters to collapse/show, clear and sort filters alphabetically and numerically.
 Each filter applied will automatically update the values in the other filter panes and will update the table data immediately.
 You can select multiple values in a filter pane by holding the [Ctrl] key on Windows or the [command] key on Mac. You can select a range of values by holding the [Shift] key on your keyboard.

Filters Active - 4 Collapse All Show All Clear All

Gene Term Types x AA⁺ #⁺ v Disease Name Types x AA⁺ #⁺ v Disease Names x AA⁺ #⁺ v

Journal Name x AA⁺ #⁺ v Publication Date x AA⁺ #⁺ v

2019 2
 2021 1
 2023 2
 2024 1
 2025 2

Q Search Results Export Table Data 25 entries per page

Gene ID	Gene Term Types	Gene Terms	Disease ID	Disease Name Types	Disease Names	Article Title & Information
<input type="checkbox"/> 3043	GENE SYMBOL	HBB	0008614 GARD	Alias	<ul style="list-style-type: none"> Scd Sickle Cell Disease 	Newborn Screening for Sickle Cell Disease and Thalassemia. Journal: JAMA health forum (2025) PMID: 40053336
<input type="checkbox"/> 3043	GENE SYMBOL	HBB	0008614 GARD	Alias	Scd	New Born Screening of Hemoglobinopathies in a Center Tunisian Population. Journal: Journal of pediatric hematology/oncology (2024) PMID: 38748601
<input type="checkbox"/> 3043			0008614		Scd	Scalable noninvasive amplicon-based precision sequencing (SNAPseq) for genetic diagnosis and screening of β-thalassemia and sickle cell disease

PubMed User Guide

[JAMA Health Forum](#). 2025 Mar 7;6(3):e250064. doi: 10.1001/jamahealthforum.2025.0064.

Newborn Screening for Sickle Cell Disease and Thalassemia

Maa-Ohui Quarmyne¹, Fiona Bock², Sangeetha Lakshmanan², Brandon K Attell², Angela Snyder², Jeanne Boudreaux³, Sujit Sheth⁴, M A Bender⁵, Ashutosh Lal⁶

Affiliations [+ expand](#)
 PMID: 40053336 DOI: 10.1001/jamahealthforum.2025.0064 [Free article](#)

Abstract

Importance: Hemoglobin disorders are a considerable public health issue with more than 500 000 affected infants born annually worldwide. First introduced in the 1970s, newborn screening (NBS) for sickle cell disease (SCD) was included in the Recommended Uniform Screening Panel (RUSP) in 2006, a successful public health promotion and prevention practice that has led to improved childhood survival. Although SCD is the primary target, the screening process also detects many other hemoglobinopathies.

Observations: NBS programs, administered by individual states, vary in their practices for hemoglobinopathy screening, creating health inequities and compromising public health efforts. There is a lack of uniformity in the choice of primary screening test, reporting, and follow-up of abnormal results, exacerbated by inconsistent access to genetic confirmation. Consequently, newborns diagnosed through protein-based screening alone may have diverse genotypes that alter the clinical expression of hemoglobinopathies. This Special Communication considers how the universal adoption of molecular testing for hemoglobinopathy newborn screening can overcome these current shortcomings. Simultaneously, the considerable challenges of primary screening with molecular methods and how these can be overcome are evaluated. Screening with targeted genetic

FULL TEXT LINKS
[JAMA Health Forum](#) [FULL TEXT](#)

ACTIONS

PAGE NAVIGATION
[Title & authors](#)
[Abstract](#)
[Similar articles](#)
[Cited by](#)
[MeSH terms](#)
[Related information](#)
[LinkOut - more resources](#)

Example C: Explore LiteratureAI (Cont'd)

- 7** To start a new search or make modifications to an existing search, utilize the different tabs at the top of the page to switch between the different types of literature searches.

Literature Search Results for Gene-Disease Co-occurrence

1. Indicate whether your search will start with a gene, or with a disease
2. Type to search, or select from the list of genes or diseases provided.
3. Select one or more values from the list of all known genes/diseases related to your selection in step 2, then click "Display".

1 Start with: **Diseases** 2 Select Gene: HBB 3 Select Disease: **Hb SS disease** Display Reset

Currently displaying results for: **HBB and Hb SS disease**

Top 10 Journals by Publication Count

Journal	PubMed Articles
Hemoglobin	~48
Blood	~25
British journal of haematology	~15
The Cochrane database of systematic reviews	~12
PloS one	~10
American journal of hematology	~8
National Academy of Sciences of the United States of America	~6
Blood cells, molecules & diseases	~5
Clinical chemistry	~4
International journal of molecular sciences	~3

Number of Publications by Year (All Journals)

Year	PubMed Articles
1974	~1
1975	~1
1976	~1
1977	~1
1978	~1
1979	~1
1980	~1
1981	~1
1982	~1
1983	~1
1984	~1
1985	~1
1986	~1
1987	~1
1988	~1
1989	~1
1990	~1
1991	~1
1992	~1
1993	~1
1994	~1
1995	~1
1996	~1
1997	~1
1998	~1
1999	~1
2000	~1
2001	~1
2002	~1
2003	~1
2004	~1
2005	~1
2006	~1
2007	~1
2008	~1
2009	~1
2010	~1
2011	~1
2012	~1
2013	~1
2014	~1
2015	~1
2016	~1
2017	~1
2018	~1
2019	~1
2020	~1
2021	~1
2022	~1
2023	~1
2024	~1
2025	~1

- 8** Repeat steps 3 through 6 to continue exploring articles and diving further into the publications. To view the comprehensive LiteratureAI features, see the next page.

Literature Search Results for Diseases

Please search for a disease from the dropdown below and then select from the list of values provided.

Clicking "See Disease-Genes Co-occurrence" will display literature for the disease and all known associated genes.

Select a Disease: **Cystic fibrosis** GARD Disease ID: 0006233 See Disease-Genes Co-occurrence →

Top 10 Journals by Publication Count

Journal	PubMed Articles
fibrosis: official journal of the European Cystic Fibrosis Society	~2800
Pediatric pulmonology	~1800
American journal of respiratory and critical care medicine	~1200
PloS one	~1000
The Journal of pediatrics	~800
The European respiratory journal	~700
Thorax	~600
The Journal of biological chemistry	~500
Chest	~400
Lancet (London, England)	~300

Number of Publications by Year (All Journals)

Year	PubMed Articles
1974	~10
1975	~10
1976	~10
1977	~10
1978	~10
1979	~10
1980	~10
1981	~10
1982	~10
1983	~10
1984	~10
1985	~10
1986	~10
1987	~10
1988	~10
1989	~10
1990	~10
1991	~10
1992	~10
1993	~10
1994	~10
1995	~10
1996	~10
1997	~10
1998	~10
1999	~10
2000	~10
2001	~10
2002	~10
2003	~10
2004	~10
2005	~10
2006	~10
2007	~10
2008	~10
2009	~10
2010	~10
2011	~10
2012	~10
2013	~10
2014	~10
2015	~10
2016	~10
2017	~10
2018	~10
2019	~10
2020	~10
2021	~10
2022	~10
2023	~10
2024	~10
2025	~10

Example C: Explore LiteratureAI (Cont'd)

Browse Literature Associated With Genes & Rare Diseases

Rare disease and genes are continuously being added to LitAI as our efforts are fully aligned and coordinated with ongoing GARD updates.

Genes – literature search results for gene mentions alone
 Diseases – literature search results for disease mentions alone
 Gene-Disease Co-occurrence – literature search results where both the rare disease and associated gene are mentioned. Results in the 'Gene-Disease Co-occurrence' category are limited as the feature looks for both gene and disease mentions. We recommend 'Diseases' and 'Genes' tabs for finding rare disease related articles of interest.

Notifications & Updates

- Literature AI now includes latest MEDLINE information up to Apr 03, 2026.
- Currently, we provide results for diseases with known gene associations, and we encourage you to periodically check-in as we make progress to incorporate all known rare diseases.
- Please [contact us](#) if you do not find a particular rare genetic disease, or if there are issues with the current results.

1 Genes Diseases Gene-Disease Co-occurrence

Literature Search Results for Genes

Q Please select a gene from the dropdown below to perform your search. You can type to search for a specific gene, or select one from the list provided.
 ⓘ Clicking "See Gene-Disease Co-occurrence" will display literature for the gene and all known associated diseases.

Select a Gene: **2** CPS1 **3** Carbamoyl-phosphate synthase 1 **4** Entrez Gene ID: 1372 See Gene-Disease Co-occurrence →

5 Top 10 Journals by Publication Count

Journal	Publication Count
Scientific reports	18
PLoS one	14
The Journal of biological chemistry	14
Molecular genetics and metabolism	12
Journal of bacteriology	11
International journal of molecular sciences	10
Human mutation	9
Journal of inherited metabolic disease	8
Molecular genetics and metabolism reports	7
Hepatology (Baltimore, Md.)	6

6 Number of Publications by Year (All Journals)

1 Category Tabs
Switch between LiteratureAI disease, gene, or gene-disease co-occurrence.

2 Search Box
Search for a disease, gene, or gene-disease co-occurrence.

3 Disease Information
Displays disease, gene, or disease-gene details.

4 Gene-Disease Co-occurrence Button
In disease or gene mode, quickly swap to co-occurrence search.

5 Top Publication Journals
Bar chart showing the top 10 journals by publication count.

6 Publication Trends by Year
Line graph tracking research output over time.

Example C: Explore LiteratureAI (Cont'd)

1 Filters Active - 0

2 Search Results:

3 Collapse All Show All Clear All

4 Types Gene Terms Journal Name Publication Date

5 Export Table Data

6 Comprehensive transcriptomic analysis reveals canonical and novel pathways modulated by nanocarta in mammalian retinal degeneration.

Journal: Scientific reports (2020)
PMID: 41494209

Targeting CP 51 attenuates lung cancer metastasis by regulating EMT through an epigenetic mechanism.

Journal: Theranostics (2020)
PMID: 41356169

Corrigendum to "CircSETD2 impairs hepatic lipid homeostasis in melastotic dysfunction-associated fatty liver disease by binding CP51" [Int. J. Biol. Macromol. Volume (year) start page - end page, Reference: BIOMAC 145575, PII: S0141-8130(20)29456-X].

Journal: International journal of biological macromolecules (2020)
PMID: 41344936

Pulmonary Solid and Granular Adenocarcinomas Expressing HepPar1/CP51: Highly Aggressive Tumors Exhibiting Mitochondrial Adaptation to STK11 Mutations Rather Than Hepatoid Differentiation.

Journal: Modern pathology : an official journal of the United States and Canadian Academy of Pathology, Inc (2020)
PMID: 41580209

Neurotransmitter receptor-associated gene signature: prognostic and immunosuppressive microenvironment in NSCLC.

Journal: Future science OA (2020)
PMID: 41492772

Carbon starvation induces coincident capsule and cell wall remodeling in *Cryptococcus neoformans*.

Showing 1 to 25 of 567 entries

- 1 Filtering**
Narrow results to main terms, aliases, journals, and publication dates.
- 2 Keyword Search**
Use keywords to match terms from article titles, journal names, and PMIDs.
- 3 View Settings**
Use button control to collapse/show, and clear filters.
- 4 Advance Filtering**
Filter alphabetically or numerically, hold ctrl key for multiple selections.
- 5 Export Result Table**
Download detailed comma separated values file of all results and corresponding annotations.
- 6 PubMed Access**
Click article title to be directed to full PubMed publication.

Conclusion

RARe-SOURCE®, an integrated bioinformatics resource for rare diseases, is designed to help researchers and clinicians uncover insights into rare diseases. Data from multiple sources are reviewed, annotated, integrated and presented to facilitate discoveries into rare disease therapeutics.



Visit <https://raresource.nih.gov/> to learn more