

Breakout Session 1: Track A

**Rare Disease Alert System
(RDAS)**

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Team Lead, NIH/NCATS

NCATS

COLLABORATE. INNOVATE. ACCELERATE.

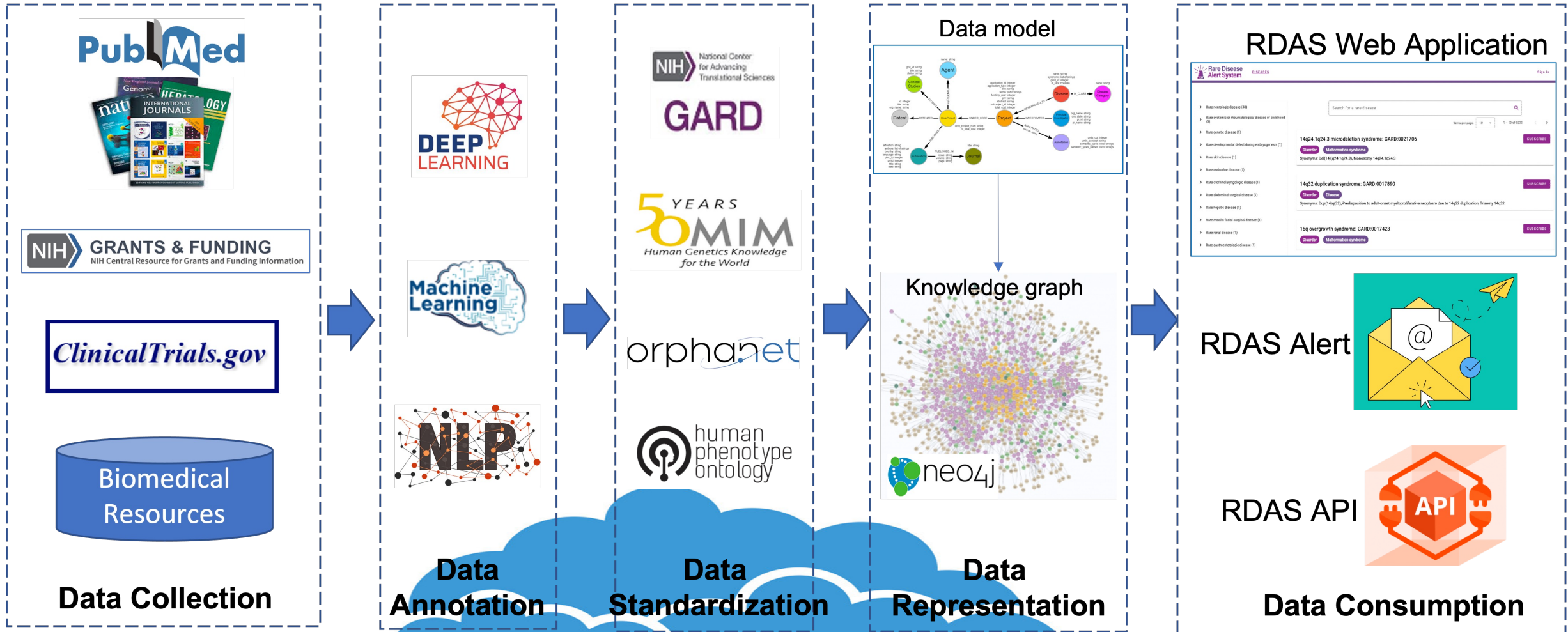
Rare Disease Alert System (RDAS)

Qian Zhu, PhD
IFX/DPI/NCATS
1/17/2024

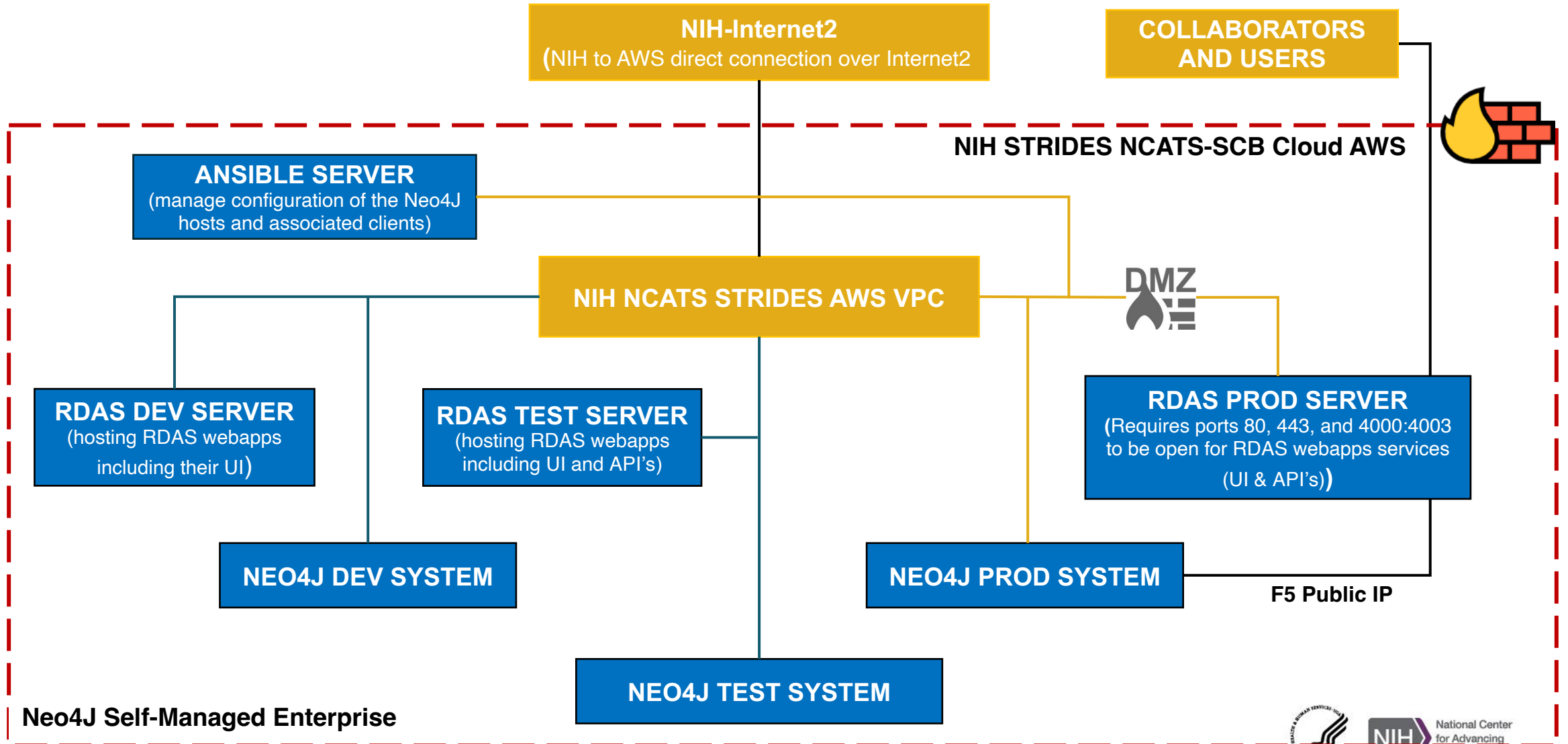


NIH National Center
for Advancing
Translational Sciences

Rare Disease Alert System



RDAS STRIDES Network Infrastructure



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



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 UI – Disease page

The screenshot displays the RDAS UI interface for a disease page. The top navigation bar includes the 'Rare Disease Alert System' logo, 'DISEASES', 'ABOUT', 'APIs', and a 'Sign In' link. The main content area is divided into a left sidebar and a main panel. The sidebar contains 'Filters' and 'Hierarchy' sections. The 'Filters' section has a search box for phenotypes and a list of phenotypes with checkboxes and counts: Seizure (1345), Intellectual disability (1223), Global developmental delay (1140), Short stature (1014), Microcephaly (857), Hypotonia (716), and Scoliosis (704). The 'Hierarchy' section is currently empty. The 'Genes' section has a search box. The main panel features a search bar, a 'Sort' dropdown menu set to 'Projects', and 'Items per page' set to 10. Below this, two disease entries are shown: 'Lymphoma: GARD:0020548' and 'Tuberculosis: GARD:0007827'. Each entry has a 'Sign In' link and a help icon. The 'Lymphoma' entry has filters for 'Group of disorders' and 'Category', and statistics for Published Articles (9973), Funded Projects (4944), Clinical Trials (2468), Associated Genes (0), and Phenotypes (0). The 'Tuberculosis' entry has filters for 'Disorder' and 'Disease', and statistics for Published Articles (9879), Funded Projects (3803), Clinical Trials (75), Associated Genes (1), and Phenotypes (5). The footer contains the text 'National Center for Advancing Translational Sciences'.

Rare Disease Alert System DISEASES ABOUT APIs [Sign In](#)

Rare disease search

Sort: Projects Items per page: 10 1 – 10 of 12004

Lymphoma: GARD:0020548 [Sign In](#) ?

Group of disorders Category

Published Articles	Funded Projects	Clinical Trials	Associated Genes	Phenotypes
9973	4944	2468	0	0

Tuberculosis: GARD:0007827 [Sign In](#) ?

Disorder Disease

Published Articles	Funded Projects	Clinical Trials	Associated Genes	Phenotypes
9879	3803	75	1	5

National Center for Advancing Translational Sciences

Chronic myeloid leukemia and monitoring

American journal of hematology

Kantarjian H, Jabbour A, Kantarjian H, et al. Department of Leukemia, University of Texas MD Anderson Cancer Center, Houston, Texas, U.S.A.

STUDIES ON THE DEVELOPMENT OF

Abstract

We wish to test the hypothesis that the bone marrow. This idea was based on the findings of previous studies of lymphomas arise in the lymph nodes. In the present study, we used hybridization techniques to identify determinants expressed by

Phase II Trial of Sequential Chemotherapy and Radiotherapy for AIDS-Related Primary Central Nervous System Lymphoma

[NCT00000801](#)

Study Type

Interventional

Status

Completed

Phase

Phase 2

Summary

To estimate the response rate, overall and disease-free survival, toxicities, factors associated with outcome, and effect on quality of life in patients with AIDS-related primary CNS lymphoma treated with CHOD (cyclophosphamide, doxorubicin, vincristine, and dexamethasone) plus filgrastim (granulocyte-colony stimulating factor; G-CSF) and external beam irradiation. To determine other clinical markers present in this patient population. Combined modality therapy may prove of benefit for patients with AIDS-related primary CNS lymphoma.

First Posted

Unknown

Last Update

November 1, 2012

Epidemiologies

Epidemiology and incidence
Epidemiology
1-2 cases per 100,000
Location:
United States

Funding

Funding Agency

National Cancer Institute

Total Cost

\$179,272.00

Sub Projects: 3

< Study Design Participant Info Study Diseases Conditions (2) Locations (7) Sponsor >

Show Abstract

1987 STUDIES ON THE DEVELOPMENT OF

Substances

Imatinib Mesylate

Diseases Studied

[Lymphoma](#)

Design 1

Primary Purpose

Treatment

Detailed Description

Combined modality therapy may

Other Diseases

[Chronic myeloid leukemia](#)

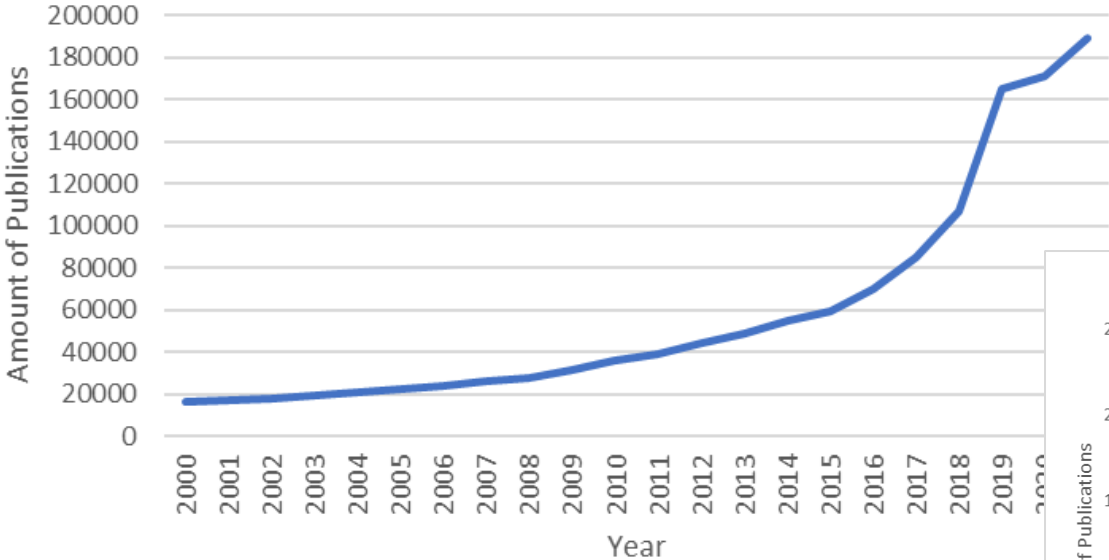
RDAS neo4j - https://rdas.ncats.nih.gov/browser

The screenshot displays the RDAS neo4j browser interface. The browser address bar shows the URL `rdas.ncats.nih.gov/browser/?iss...`. The interface is divided into several sections:

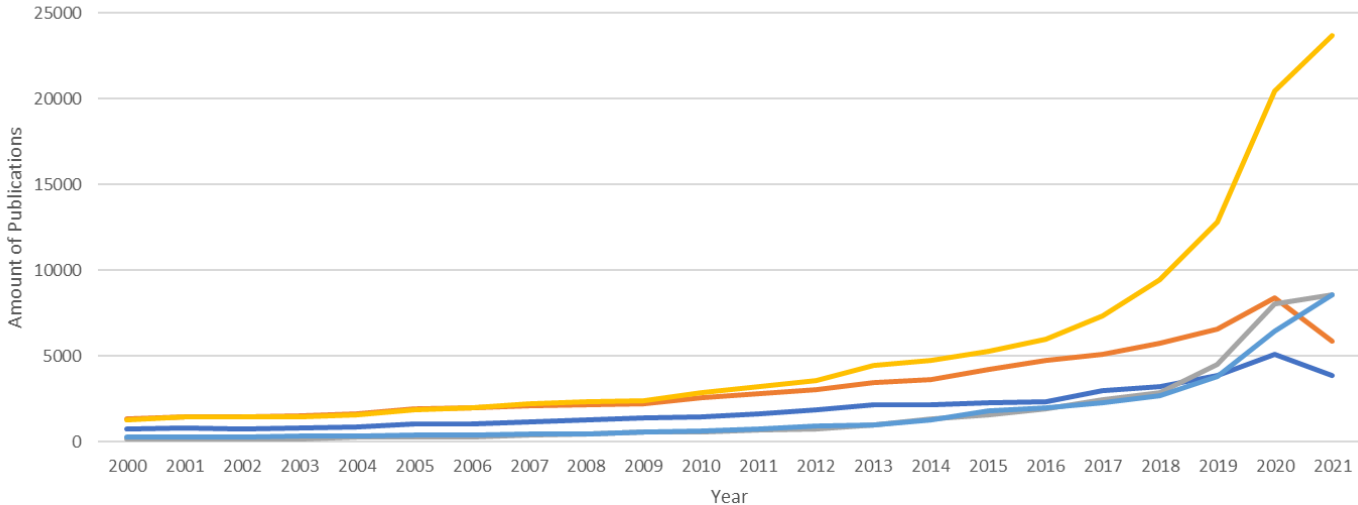
- Left Sidebar:** Contains a search bar with a dropdown menu showing options like "clinical", "gard", "grant", "neo4j", "pubmed" (selected), and "system". Below this are "Node labels" and "Relationship types" sections, each with a list of filter buttons and their associated counts.
- APIs Menu:** A vertical menu on the left side of the main content area, listing categories: "APIs", "EPIDEMIOLOGY", "DISEASES", "PUBLICATIONS", "PROJECTS", and "TRIALS".
- Main Content Area:** Displays a graph visualization with a central node labeled "Severe renal disease" and numerous surrounding nodes connected by edges. A query editor at the top of this area shows a Cypher query: `... ticle) RETURN n LIMIT 25`.
- Node Properties Panel:** Located on the right side, it shows details for the selected node, including its ID (552), creation date (12/08/22), and abstract text: "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...".

RD based publication landscape analysis

Number of publications per year



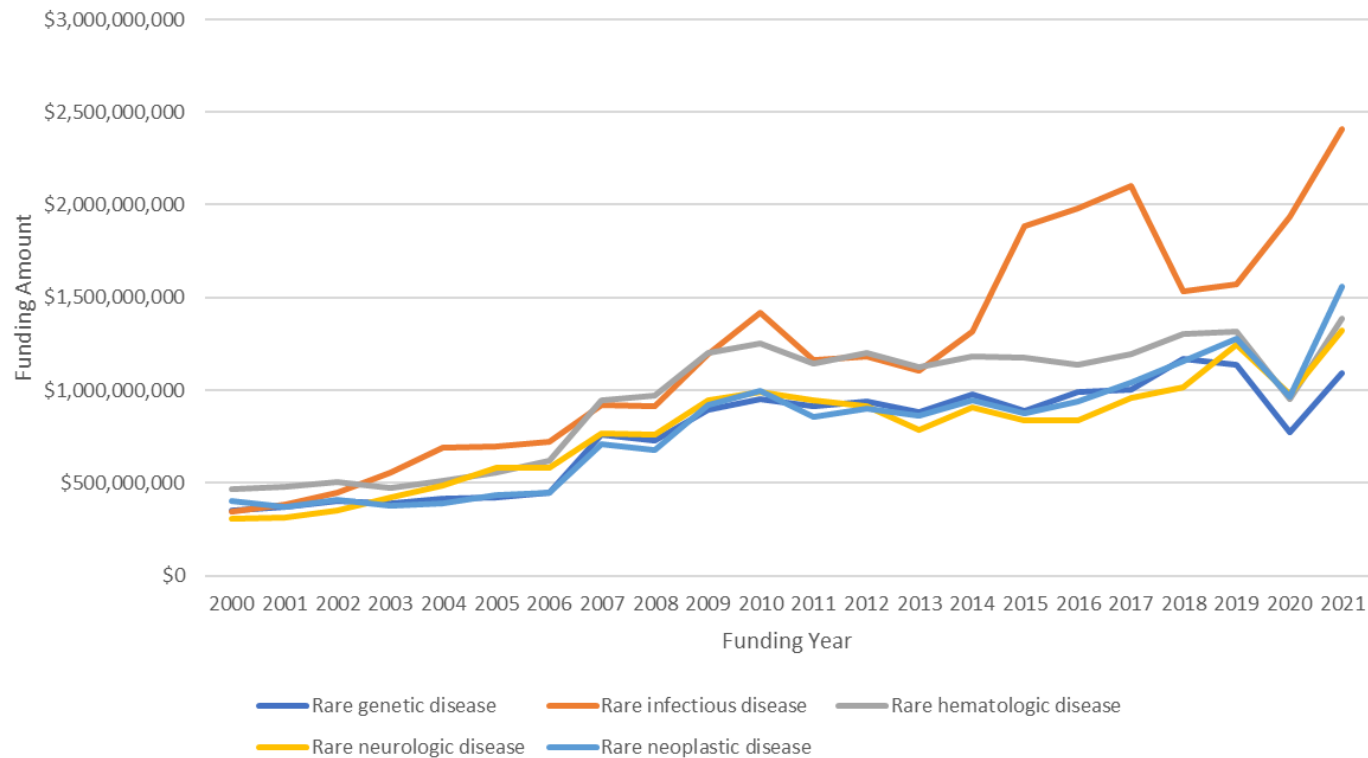
Number of Publications for Rare Disease Categories per Year



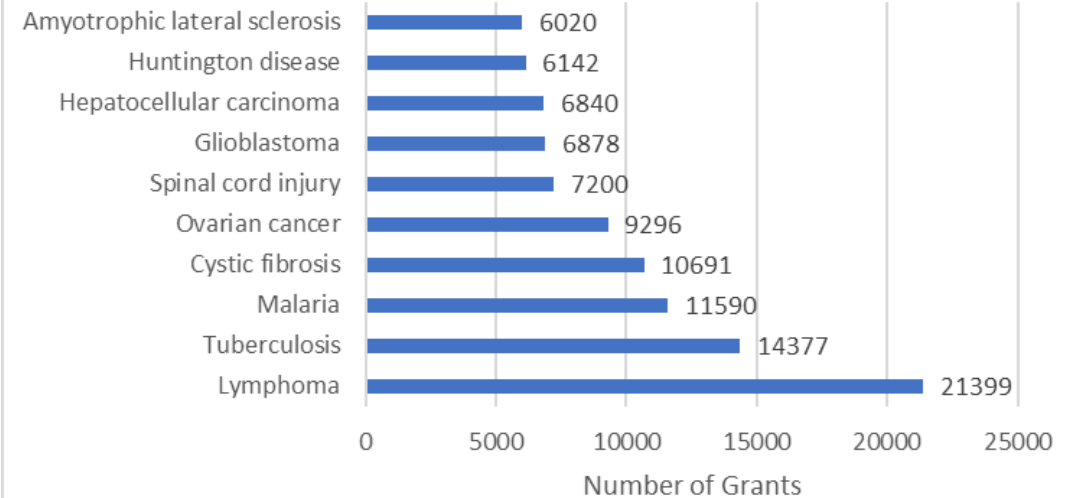
- Rare developmental defect during embryogenesis
- Rare genetic disease
- Rare infectious disease
- Rare neoplastic disease
- Rare disorder potentially indicated for transplant or complication after transplantation

RD based funding landscape analysis

Total Funding for Rare Disease Categories per Year



Top 10 GARD Rare Diseases



RD based CT landscape analysis

Number of clinical trials by intervention types

Intervention Type	# CT
Drug	123,863
Other	32,421
Device	15,273
Procedure	19,143
Biological	18,077
Behavioral	9,297
Diagnostic Test	5,549
Radiation	4,520
Dietary Supplement	4,176
Genetic	1,945
Combination Product	838

Top 10 rare diseases with the greatest number of clinical trials

GARD ID	GARD Name	# CT
GARD:0020511	Rare carcinoma of pancreas	4069
GARD:0021758	Hereditary gastric cancer	3412
GARD:0006383	Carcinoma of esophagus	3222
GARD:0007108	Multiple myeloma	2718
GARD:0020548	Lymphoma	2594
	Malignant lymphoma with peripheral neuropathy	
GARD:0020387		2592
GARD:0021062	Primary oculocerebral lymphoma	2592
GARD:0021064	Primary organ-specific lymphoma	2592
GARD:0020110	Malignant melanoma of the mucosa	2320
GARD:0012772	Rare malignant breast tumor	2294



Acknowledgement

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Timothy Sheils



Sungrim Moon

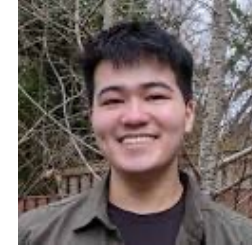


Ewy A. Mathé

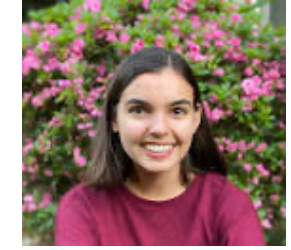
Previous IFX trainees



William Kariampuzha



Jeremy Parker Yang



Jennifer John

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Devon Leadman



Jaber Valinejad



Carlin Biyoo



Hugo Hernandez



Mariam Deacy



Sam Michael

DRDRI/NCATS



Yanji Xu

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