

Getting Ready for Research as a Rare Disease Patient Advocate

Christopher P. Austin, M.D.
Director, NCATS

NORD RareLaunch Research Ready Workshop
December 3, 2020

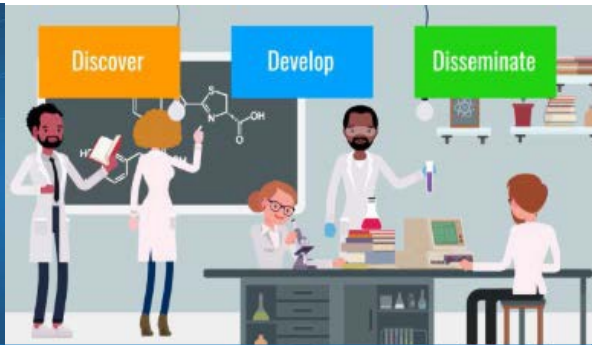


Changing the Narrative: *Rare Diseases Are a Public Health Issue*

- ~7000 diseases
 - ~80% genetic
 - ~60% onset in childhood
 - ~250 new rare diseases identified every year
- Individually rare, cumulatively common
 - Definition varies by country: US <200,000; Japan <50,000; EU <1/2,000
 - Total prevalence ~8% (US ~25 million)
- High costs in direct patient care, loss of productivity
- Accurate diagnosis often requires 5-15 yrs
- Only 5% of rare diseases have a regulatorily approved treatment
 - ~400 diseases; at current rate 2000 years before treatments for all rare diseases
- Solution: transition from “one disease at a time” to “many diseases at a time” approach
 - Commonalities among diseases
 - Platform technologies for diagnosis and treatment



NCATS Office of Rare Diseases Research



Knowledge/Data	Research/Collaboration	Community Engagement
GARD Cluster	Rare Diseases Clinical Research Network	Rare Diseases Day at NIH
Genetics & Rare Diseases (GARD) Information Center		Rare Diseases Are Not Rare! Challenge
Rare Diseases Registry Program (RaDaR)	NCATS Scientific Conference Grants	Gene Therapy Meeting Series
NCATS Toolkit for Patient-focused Drug Development	Bench to Bedside Grants	
Impact of Rare	Clinical Trial Readiness Grants	International Collaboration IRDiRC
Diagnostic Odyssey	PaVe-GT	
	Shared Molecular Etiologies Grants	



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 Director
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NCATS “Many Diseases at a Time” Research Programs

Providing Information and “Big Data”

NIH National Center for Advancing Translational Sciences | **GARD** Genetic and Rare Diseases Information Center | 1-888-205-2311

Diseases | Guides | News | About GARD | En Español

GARD Information Specialists can provide you with current, reliable, and easy to understand information about rare or genetic diseases in English or Spanish.

Browse Diseases A to Z
Find Advocacy Organizations
Search GARD Glossary
Find Tools To Speed Research

Search for Diseases, Organizations, News and More... GO

About GARD

The Genetic and Rare Diseases Information Center (GARD) is a program of the National Center for Advancing Translational Sciences (NCATS) and is funded by two parts of the National Institutes of Health (NIH): NCATS and the National Human Genome Research Institute (NHGRI). GARD provides the public with access to current, reliable, and easy-to-understand information about rare or genetic diseases in English or Spanish.

Empowering Patients as Research Partners

NIH National Center for Advancing Translational Sciences | **Toolkit** For Patient-Focused Therapy Development

Home | Getting Started | Discovery | Preparing for Clinical Trials | Clinical Trials and FDA Review | After FDA Approval | About

Working Together to Advance Rare Diseases Research

Welcome! This Toolkit was developed to provide your patient group with the tools needed to advance medical research. Our goal is to ensure that patients are engaged as essential partners from beginning to end of the research and development process. This is a living site where you will find tools being developed for and by patient groups in concert with their academic, government, industry and advocacy partners. Read more about why NCATS developed this Toolkit.

Developing Interoperable Registries

NIH National Center for Advancing Translational Sciences | **RaDaR** Rare Diseases Registry Program

About the Rare Diseases Registry Program

About the RaDaR Program

The Rare Diseases Registry (RaDaR) Program, formerly known as the Global Rare Diseases Registry Data Repository (GRDR) program, aims to provide easily accessible advice for constructing and maintaining good-quality rare disease patient registries to enable therapeutics development.

The RaDaR website is under construction. Please sign up below or check back for new information as the site is updated.

Natural History and Interventional Studies



National Center for Advancing Translational Sciences

NCATS “Many Diseases at a Time” Research Programs

Partnering to Change Perceptions



Challenges About Agency Toolkit Contact

NCATS Rare Diseases Are Not Rare! Challenge

Share innovative ways to communicate to educate people about rare diseases through social media or art

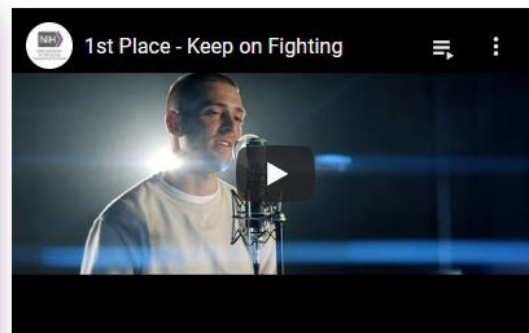
First Place 2018
Nancy Netherland

First Place 2020

“Keep on Fighting”

Jacob Thompson

“When I was 24 years old, about 2 years ago, I was diagnosed with the rare disease known as Friedreich’s Ataxia (FA). When my dreams of playing a sport in college ended, I needed something else to apply myself to. This began my journey as a hip-hop and spoken word artist. When I was diagnosed, it felt as though my dreams for the future had died. In many instances, I felt like giving up on my journey as an artist. Instead, I now have a new vision for my art: both to encourage those who are challenged, whether with rare disease or in general, and to raise awareness for rare disease.”

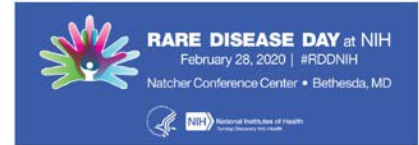


Partnering to Share Knowledge



Rare Disease Day at NIH 2020
February 28, 2020 | #RDDNIH
Natcher Conference Center • Bethesda, MD

- Generate the full commitment to helping people with rare disease through research.
- Highlight NIH-supported rare diseases research and the development of diagnostics and treatments.
- Initiate a mutually beneficial dialogue among public and private researchers, patients, patient advocates and policymakers.
- Advance the latest rare disease information with stakeholders to advance research and therapeutic efforts.
- Put a face on rare diseases by sharing stories of patients, their families and their communities.



NCATS, FDA Co-Host Workshop on Gene Therapy for Rare Diseases

Translational Science Highlight

- NCATS and the Food and Drug Administration are collaborating to identify bottlenecks to gene therapy development and share best practices for advancing gene therapies, a promising approach to treating many different rare diseases.

“We are now at the point of translating the potential of gene therapy — which has been around for a while — into a reality for patients,” said NCATS Director Christopher P. Austin, M.D. These new advances in gene therapies have truly been a long time in coming. The first human study that involved gene therapy, a type of treatment to replace or fix a defective gene in a patient, began in 1990 at the NIH Clinical Center. Nearly 30 years later, in 2017, the Food and Drug Administration (FDA) approved the first three gene therapies for use in the U.S. One of those three is for an inherited disease; the others alter a patient’s own cells to fight two forms of cancer.

Inspired by this scientific progress, in August 2018, NCATS and the FDA co-hosted a **two-day workshop** about gene therapy for rare diseases. Many of these diseases are caused by a mutation in a single gene, so gene therapy holds great promise as an effective treatment approach.

Led by NCATS’ Office of Rare Diseases Research and the FDA’s Center for Biologics Evaluation and Research (CBER) staff, the workshop brought together representatives from NIH, the FDA, academia, industry and patient organizations to discuss using gene therapy to treat rare diseases. Discussion topics included recent scientific advances, challenges in manufacturing the viruses, called vectors, that are used to deliver gene therapy products; and how to increase the number of available treatments.

“We really are having this meeting at the right time,” said FDA CBER Director Peter Marks, M.D., Ph.D. He noted that hundreds of potential gene therapy treatments are now entering the clinic for testing in people. In July 2018, the FDA released draft guidance on several topics related to gene therapy, such as manufacturing and long-term follow-up of people who receive these agents.

Advancing the Science

Exciting scientific advances discussed at the meeting included research in animal models and studies showing new advances in humans.

Jay Chiorini, Ph.D., of the NIH National Institute of Dental and Craniofacial Research, shared his research on using gene therapy to make salivary glands work better. Most people who have radiation treatment for head and neck cancer temporarily or permanently lose the ability to make saliva. Without it, eating becomes painful, and people may lose their teeth and have other dental problems. Using gene therapy in mini-pigs that were treated with radiation, Chiorini has been able to show some improvement in how the salivary glands worked. This approach has now entered the clinic, and he has so far enrolled 12 people in a study designed to test different gene therapy doses to find out whether the approach is safe in humans.

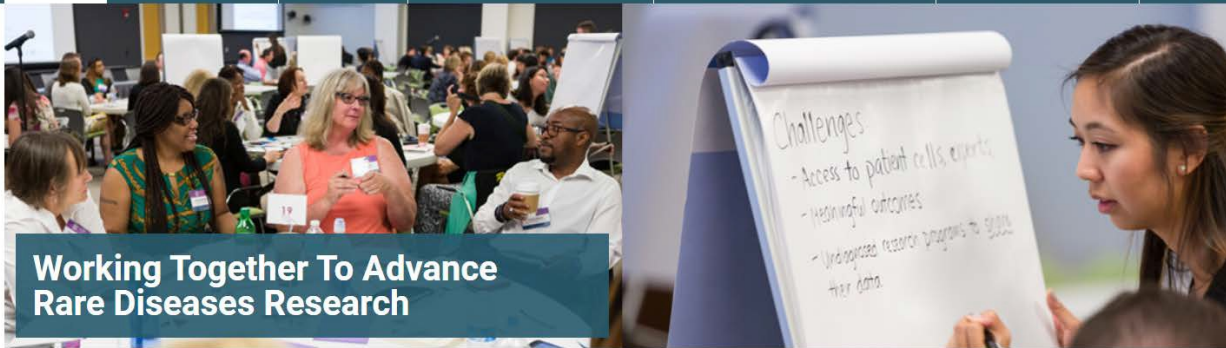
“We are often faced with the fact that mouse studies don’t



NCATS Director Christopher P. Austin, M.D., at the Growing Promise of Gene Therapy Approaches to Rare Diseases Workshop in August 2018.



NIH National Center for Advancing Translational Sciences



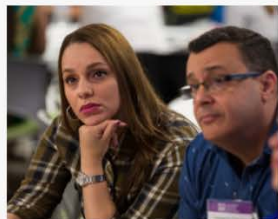
Working Together To Advance Rare Diseases Research

The National Center for Advancing Translational Sciences (NCATS) Toolkit for **Patient**-Focused Therapy Development provides information and resources to help **patient** groups support the process of developing a treatment or cure for their disease(s). The goal is to ensure that **patient** groups are engaged as essential partners from the beginning to the end of the therapy research and development (R&D) process, whether the therapeutic approach is a **drug**, **biologic**, or **medical device**.

The information in NCATS Toolkit highlights tips and strategies from the collected resources and from presentations by representatives of **patient** groups, academia, government, and industry at conferences and public meetings, as well as informal conversations with these same stakeholders.

This is a living site. New resources and information will be added as they are developed for and by **patient** groups in concert with their academic, government, industry, and advocacy partners. You are encouraged to share any helpful resources, tips, or strategies for possible addition to NCATS Toolkit through the [Contact Page](#).

Getting Started



The information and resources in this section can help you learn about how therapies (**drugs**, **biologics**, and **medical devices**) are developed, why it is important for patients to engage throughout the process, and how to build relationships with key partners.

Getting Started

NCATS Toolkit for Patient-Focused Therapy Development

- One-stop shop for tools and advice for advancing research programs
- Brings together resources from many organizations
- Developed with and for patient groups wanting to get started in the research process

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



There Is Power in Numbers

Each person's story matters. Registries turn each patient's experience into quality data that can lead to new discoveries and research advances.

Rare Diseases Registry Program (RaDaR) Helps You:

Get Started

Learn about the different types of registries and the major milestones for building a successful registry.

Get Started

Set Up Your Registry

Create your registry plan and incorporate data standards and best practices when building your registry.

Set Up Your Registry

Manage Your Registry

Get tools and resources to recruit participants, maintain quality data, and improve your registry.

Manage Your Registry

Registries Connect Patient Communities with Researchers

A **registry** is a collection of standardized information about a group of individuals, such as those living with the same disease, that is used for a variety of specific purposes. A **registry** is a powerful tool for collecting information about patients diagnosed with the same rare disease. One difficulty of learning more about rare diseases is the challenge of finding enough people eligible to participate in research studies. The more participants in a study, the more powerful the results. Every person matters!

When you create a rare disease **registry**, you bring together the entire community, which includes patients, caregivers, clinicians, researchers, and industry. Understanding the varied needs of all members of the rare disease community is important so you can create a **registry** that patients and caregivers want to join, and clinicians, researchers, and industry want to use.

RaDaR provides step-by-step guidance and tools for building registries based on good data practices.

The Rare Diseases **Registry** Program (RaDaR) website was created to provide advice on setting up and maintaining good-quality registries for rare diseases to stimulate research. RaDaR enables collaborative sharing of information and tools to promote **data standardization** and integration from the earliest stages of **registry** development.



About RaDaR

NCATS RaDaR (Rare Diseases Registry) Program

- Tools and advice for building patient registries
- Developed with and for patient groups wanting to get started in developing a registry and natural history study

<https://registries.ncats.nih.gov/>



Diseases Guides News About GARD En Español

GARD Information Specialists can provide you with current, reliable, and easy to understand information about rare or genetic diseases in English or Spanish.



Search for Diseases, Organizations, News and More... GO

- Browse Diseases**
View diseases by alphabetical order
- Find Support**
Search for advocacy organizations
- Search GARD Glossary**
Learn about medical and genetics terms

✉️ Facebook Twitter

About GARD

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Read more [about GARD](#).

Find Out How GARD Information Specialists Can Help You



Patients, Families and Friends



Healthcare Professionals



Researchers



Teachers and Students

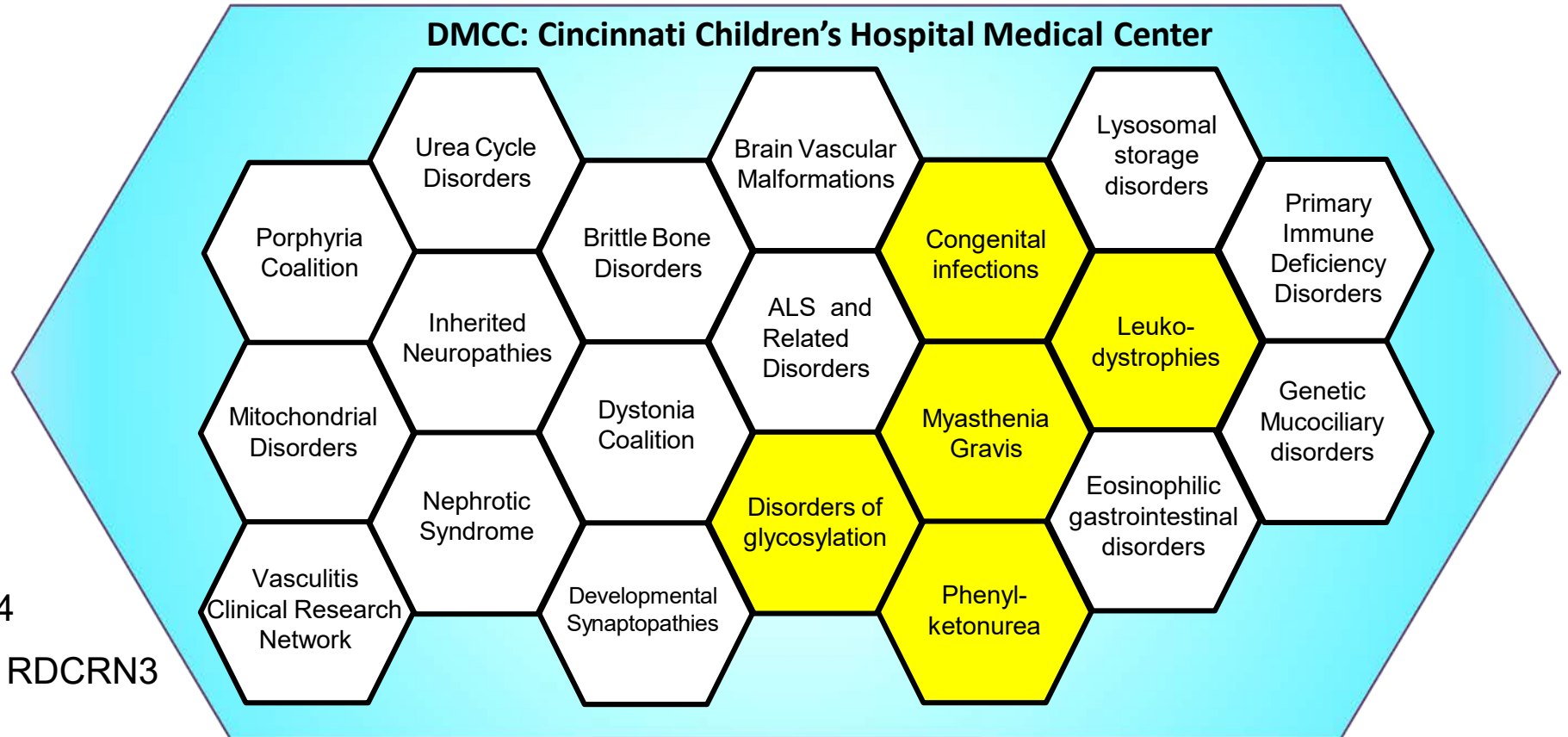
Genetic and Rare Diseases Information Center (GARD)

- Since 2003 the preeminent source of patient-focused information on all rare diseases
- Includes web-based information and consultation phone line
- Now being completely redesigned with new IT and informatics capabilities

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

- Network of consortia grouped around ≥ 3 rare diseases
- Natural history, recruitment, intervention trials required
- *Patient advocacy group inclusion required*

NIH Partners:
NCATS, NINDS, NICHD,
NHLBI, NIMH, NIDDK, NIDCR,
NIAMS, NIAID, ODS



Patient Organizations



Patient Organizations (Patient Advocacy Groups)

Patient Advocacy Groups (PAGs) are organizations that promote the needs and priorities of patients. This could include supporting research for a specific disease, promoting awareness of a disease, and educating the community about a disease, among other activities. Most patient advocacy groups represent one or a few specific diseases.

One feature of the RDCRN is the direct involvement of PAGs in network operations, activities, and strategy. Each rare disease research group (consortium) in the network includes relevant PAGs in consortium membership and activities. These PAG representatives advise the researchers within their consortium by joining network-level discussions and meetings. The RDCRN PAGs consist of PAGs directly affiliated with an RDCRN consortium, and collectively represent the perspective and interests of patients with rare diseases.

How do I find a patient organization if I don't know the rare diseases research group?

You can find out the rare diseases research group with our rare disease search tool, which will also provide you a list of the available patient organizations.

Click on a consortium name to view supporting patient advocacy groups

 Brain Vascular Malformation Consortium (BVMC)	 Brittle Bone Disorders (BBD)	 Congenital and Perinatal Infections Consortium (CPIC)	 Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR)
 CRATE: Clinical Research in ALS and Related Disorders for Therapeutic Development Consortium	 Developmental Synaptopathies Consortium (DSC)	 Dystonia Coalition (DC)	 Frontiers in Congenital Disorders of Glycosylation (FCDOG)
 Genetic Disorders of Mucociliary Clearance Consortium (GDMCC)	 Global Leukodystrophy Initiative Clinical Trials Network (GLIA-CTN)	 Inherited Neuropathy Consortium (INC)	 Lysosomal Disease Network (LDN)
 Myasthenia Gravis Rare Disease Network (MGNet)	 NEPTUNE, Nephrotic Syndrome Rare Disease Clinical Research Network	 North American Mitochondrial Diseases Consortium (NAMDC)	 Phenylalanine Families and Researchers Exploring Evidence (PHFREE)
 Porphyrias Consortium (PC)	 Primary Immune Deficiency Treatment Consortium (PIDTC)	 Rare Lung Diseases Consortium (RLDC)	 RetT Syndrome, MECP2 Duplications, & RetT-related Disorders Consortium
 STAIR: Steroid and Isoprenoid Diseases Consortium	 Urea Cycle Disorders Consortium (UCDC)	 Vasculitis Clinical Research Consortium (VCRC)	

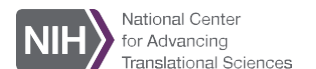
RDCRN Coalition of Patient Advocacy Groups (RDCRN-CPAG)

The RDCRN Coalition of Patient Advocacy Groups (CPAG) promotes collaboration between patient advocacy groups and the RDCRN to facilitate better access to, and earlier benefit from, research on rare diseases. As the patient advocacy arm of the RDCRN, the RDCRN-CPAG and its members use their position to advance the cause of rare diseases research and improve patient outcomes. The RDCRN-CPAG Steering Committee meets throughout the year to help facilitate the development of activities that will benefit the entire CPAG membership. Those activities include (but are not limited to) webinars, in-person meetings, and one-on-one connections with other CPAG members and RDCRN researchers.

RDCRN Coalition of Patient Advocacy Groups (CPAG)

- Integral part of RDCRN
- Information and strategy sharing among the >300 patient advocacy groups that are part of the RDCRN consortia

<https://www.rarediseasesnetwork.org/pags>



PaVe-GT: Paving the Way for Rare Disease Gene Therapies

The NCATS-led Platform Vector Gene Therapy (PaVe-GT) pilot project seeks to increase the efficiency of clinical trial startup by using the same gene delivery system and manufacturing methods for multiple rare disease gene therapies. We will make program results and regulatory documents publicly available, with the intention of benefiting future gene therapy clinical trials for very rare diseases.

WHY WE NEED PaVe-GT

Approximately 7,000 rare diseases affect 25 million people in the United States, but only about 5 percent of rare diseases have a treatment approved by the U.S. Food and Drug Administration (FDA).

Fortunately, it is becoming increasingly possible to develop gene therapies for rare diseases. A rapidly maturing therapeutic platform — adeno-associated virus (AAV) gene therapy — holds great promise for many rare diseases. AAV gene therapies use a modified version of a virus called adeno-associated virus to deliver a working copy of a defective gene into the cells relevant for a given disease. AAV has been shown to deliver therapeutic genes to cells in the eye, liver, brain, muscle and other organs. In recent years, AAV gene therapies have been approved by the FDA for treating diverse rare diseases, including a retinal disorder and a neurodegenerative condition. For many rare diseases, the limiting factor for developing a gene therapy is not scientific knowledge, but rather operational and financial hurdles, particularly for the thousands of rare diseases that are so rare there is no clear business model for bringing a gene therapy to market. These therapies for diseases of no commercial interest are sometimes referred to as “bespoke” therapies.

In theory, the process of testing new AAV gene therapies for safety prior to human trials could be streamlined because AAV gene therapies for distinct diseases can use the same outer shell, called a capsid, and the same manufacturing methods. If those two factors were kept the same, central questions in preclinical testing, like the biodistribution of a therapeutic, might need to be answered only once for each method of manufacturing an AAV gene therapy, which would save time, money and animal resources. Entire sections of Investigational New Drug (IND) applications might be able to be reused like templates. This approach may be significantly more efficient than using different manufacturing and testing processes for each gene therapy, saving precious time and resources on the road to developing and testing a gene therapy.

The goal of NCATS’ PaVe-GT pilot program is to test whether we can significantly increase the efficiency of gene therapy trial startup by using a standardized process, with the same capsid and manufacturing methods, for four different rare diseases. Importantly, we will test this hypothesis in a transparent manner so that results, documents and learnings resulting from the program will be made publicly available and can therefore be used to benefit subsequent AAV gene therapy efforts.

<https://pave-gt.ncats.nih.gov/>

LEARN MORE ABOUT PaVe-GT



Partners Across NIH: Translational Science Is a Team Sport

The PaVe-GT project is led by NCATS and includes [collaborators at different institutes](#) across NIH because surmounting obstacles in translational science requires teamwork that brings together expertise from different fields. Together, the PaVe-GT team will answer a pressing translational science question in AAV gene therapy:

Can researchers significantly increase the efficiency of gene therapy trial startup by using the same capsid and manufacturing methods for different AAV gene therapies, thereby minimizing redundancies in the preclinical development of gene therapies for extremely rare diseases?

NCATS APPROACH TO TRANSLATION

DEVELOP

NCATS designs a translational science experiment.

DEMONSTRATE

NHGRI and NINDS initiate and conduct four gene therapy programs. NCATS coordinates and harmonizes the trials.

DISSEMINATE

NCATS will share project results and documents with the public, in partnership with NHGRI and NINDS.

NCATS

NHGRI

NINDS

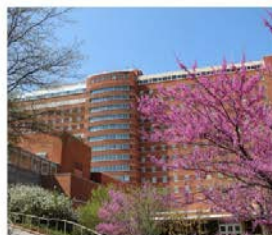
GET TO KNOW PaVe-GT



FAQS

Patient-friendly answers to top questions about PaVe-GT

LEARN MORE



DISEASES UNDER STUDY

Four rare genetic diseases will be included in the program

LEARN MORE



PUBLICATIONS

A selection of program scientific publications

LEARN MORE



RESOURCES

Gene therapy resources for researchers and patients

LEARN MORE

<https://pave-gt.ncats.nih.gov/>



NIH National Center for Advancing Translational Sciences



- News & Events
 - ▶ Releases & Announcements
 - ▶ Media Coverage
 - ▶ Grantee & Partner News
 - ▶ Events
- Stories & Publications
- Media Resources
- Connect with NCATS

NCATS is committed to including diverse patient and community perspectives in all phases of translational science, from basic research to public health implementation. Each year NCATS staff host NCATS Day to facilitate and advance robust dialogue and meaningful collaboration.

NCATS Day 2019:

NCATS DAY
2019

Conversations on Responsible Data Sharing

Date: December 17, 2019

Location: William H. Natcher Building
Natcher Conference Center
Rooms E1/E2

NCATS Day 2019, held on December 17, featured thoughtful discussion, perspectives and insights into data sharing in the translational science community.

The collection and analysis of data is important for translating research results into new prevention strategies and treatments that can improve human health. Sharing of scientific data helps validate research results, enables researchers to combine data types to strengthen analyses, facilitates reuse of hard-to-generate data or data from limited sources, and contributes to ideas for future research directions. Central to data sharing is the recognized need to make it as available as possible while ensuring the confidentiality and autonomy of research participants.

NCATS is committed to placing patients and their communities at the center of all our efforts and looks for opportunities to include the patient perspective. At NCATS Day 2019, attendees discussed data sharing from different viewpoints, including the research participant, community, researcher, federal funder and industry. Attendees taking part in this vital dialogue heard the important perspectives of stakeholders from diverse backgrounds.



Patient membership on the research team is a “disruptive technology” for rare disease therapy development

Walkley et al. *Orphanet Journal of Rare Diseases* (2016) 11:161
DOI 10.1186/s13023-016-0540-x

Orphanet Journal of
Rare Diseases

LETTER TO THE EDITOR

Open Access



Fostering collaborative research for rare genetic disease: the example of niemann-pick type C disease

Steven U. Walkley^{1*}, Cristin D. Davidson¹, Jonathan Jacoby², Philip D. Marella³, Elizabeth A. Ottinger⁴, Christopher P. Austin⁵, Forbes D. Porter⁶, Charles H. Vite⁷ and Daniel S. Ory⁸

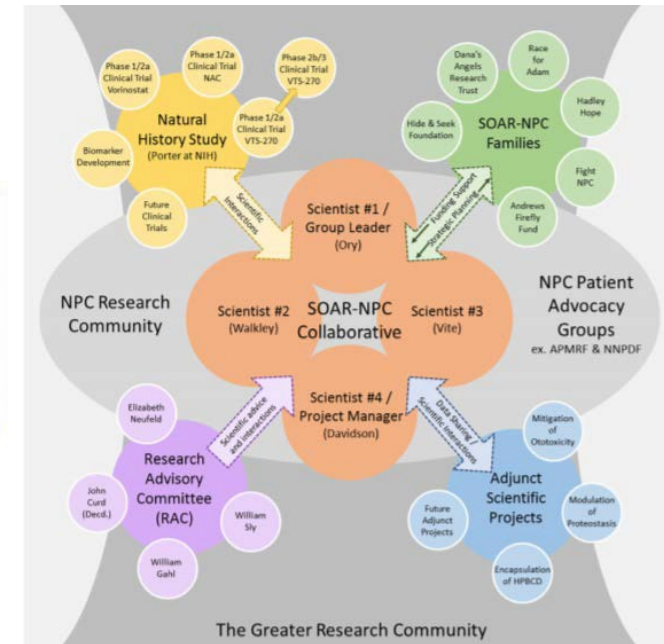
Abstract

Rare disease represents one of the most significant issues facing the medical community and health care providers worldwide, yet the majority of these disorders never emerge from their obscurity, drawing little attention from the medical community or the pharmaceutical industry. The challenge therefore is how best to mobilize rare disease stakeholders to enhance basic, translational and clinical research to advance understanding of pathogenesis and accelerate therapy development. Here we describe a rare, fatal brain disorder known as Niemann-Pick type C (NPC) and an innovative research collaborative known as Support of Accelerated Research for NPC (SOAR-NPC) which illustrates one pathway through which knowledge of a rare disease and its possible treatments are being successfully advanced. Use of the “SOAR” mechanism, we believe, offers a blueprint for similar advancement for many other rare disorders.

Keywords: Cyclodextrin, Collaborative science, Drug pipeline, Lysosomal disease, Miglustat, Niemann-Pick C, Patient advocacy, Rare disease, Therapy development, Translational medicine



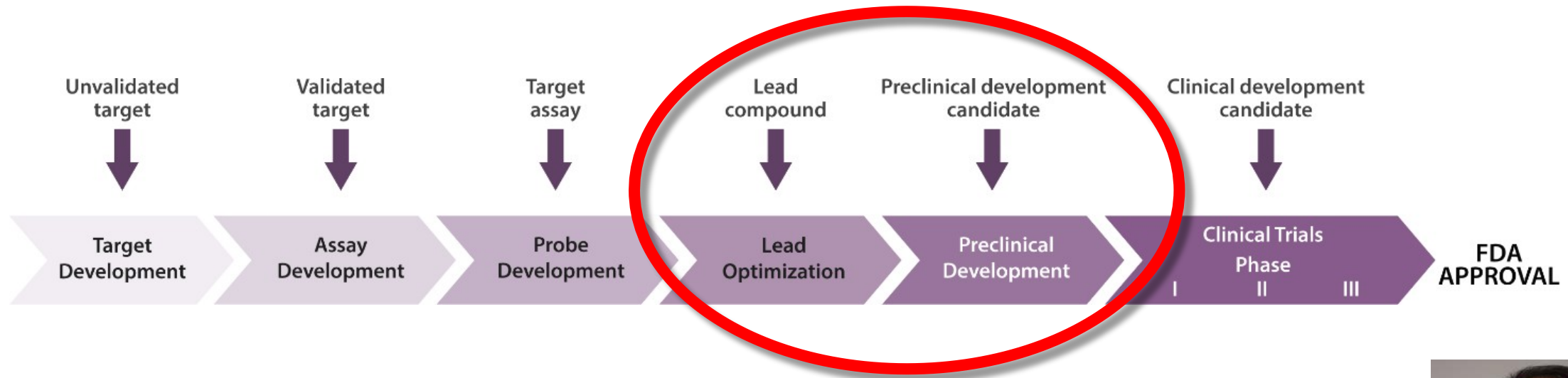
Fig. 2 SOAR-NPC Collaborative meeting. A face-to-face meeting of SOAR-NPC members as occurred at the Rose F. Kennedy Center at the Albert Einstein College of Medicine on February 26, 2014. Pictured are (left to right), Daniel S. Ory, Cristin D. Davidson, Charles H. Vite, Steven U. Walkley, Philip D. Marella and Sean Recke (not shown: Jonathan Jacoby). Individuals shown have consented to having their pictures published



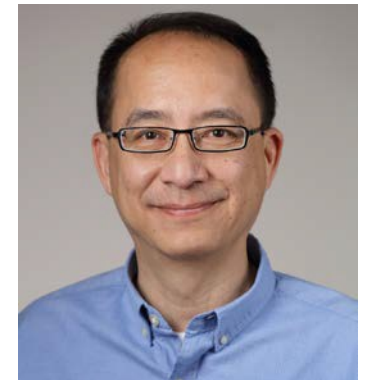
NIH National Center
for Advancing
Translational Sciences

NCATS Therapeutic Development Branch:

Lead candidate to Investigational New Drug (IND) filing with FDA



- IND clearance required to initiate clinical trials
- **IND-enabling studies** typically take 2-5 years and cost average of \$4M+
- Few funding mechanisms in this area
- Commercial investment scarce because of high failure rates
- Helping cross the drug development "**Valley of Death**" is TDB's *raison d'etre*



Don Lo, Director

<https://ncats.nih.gov/staff/lodc>

<https://ncats.nih.gov/trnd>



NIH National Center for Advancing Translational Sciences

Tissues-on-Chips

(a.k.a., “microphysiological systems”)
in vitro mimics of human organ
function



Scaffold

Cells

Structure

**Spatial and
Temporal Patterning**

Perfusion

Bioreactor

Innervation

Host Response

Functional Readout

**Computational
Design**

- Microfluidic cell culture devices

- Created with microchip manufacturing methods

- Contains continuously perfused chambers

- Seeded by human-derived cells

- Cytoarchitecture mimics tissue- and organ-level physiology

- High-resolution, real-time imaging and *in vitro* analysis of biochemical, genetic and metabolic activities



National Center
for Advancing
Translational Sciences

Using tissue chips to model diseases and drug efficacy

Kam Leong, Columbia U

Proteus Syndrome and DiGeorge Syndrome

Danielle Benoit, Lisa Delouise, Catherine Ovitt, U Rochester

Radiation-induced xerostomia

Kevin Kit Parker, William Pu, Harvard U

Barth syndrome, Catecholaminergic polymorphic ventricular tachycardia, Arrhythmogenic cardiomyopathy

Steven George, David Curiel, Stacey Rentschler, UC Davis and WashU

Atrial fibrillation

Joseph Vincent Bonventre, Luke Lee, Brigham and Women's

Polycystic kidney disease, Focal segmental glomerulosclerosis

Christopher Hughes, UC Irvine

Hereditary hemorrhagic telangiectasia, Port Wine stain, Sturge-Weber syndrome

Rocky Tuan, U Pittsburgh

Osteoarthritis, inflammatory arthritis, adipose-mediated diabetic joint complications

Clive Svendsen, Cedars-Sinai

ALS; Parkinson's Disease

Aaron Bowman, Kevin Ess, John Wiksw, Vanderbilt U

Tuberous sclerosis complex (TSC) epilepsy, DEPDC5-associated epilepsy, & associated cardiac dysfunction

Gordana Vunjak-Novakovic, Columbia U

Dox induced cardiomyopathy; multi-system pathologies involving heart, liver, skin, bone and vasculature

Donald Ingber, Harvard U

influenza infection, COPD

Jonathan Himmelfarb, U Washington

Apolipoprotein L1 mediated kidney disease, drug induced and host-pathogen interaction induced renal thrombotic microangiopathies

Teresa Woodruff, Northwestern U

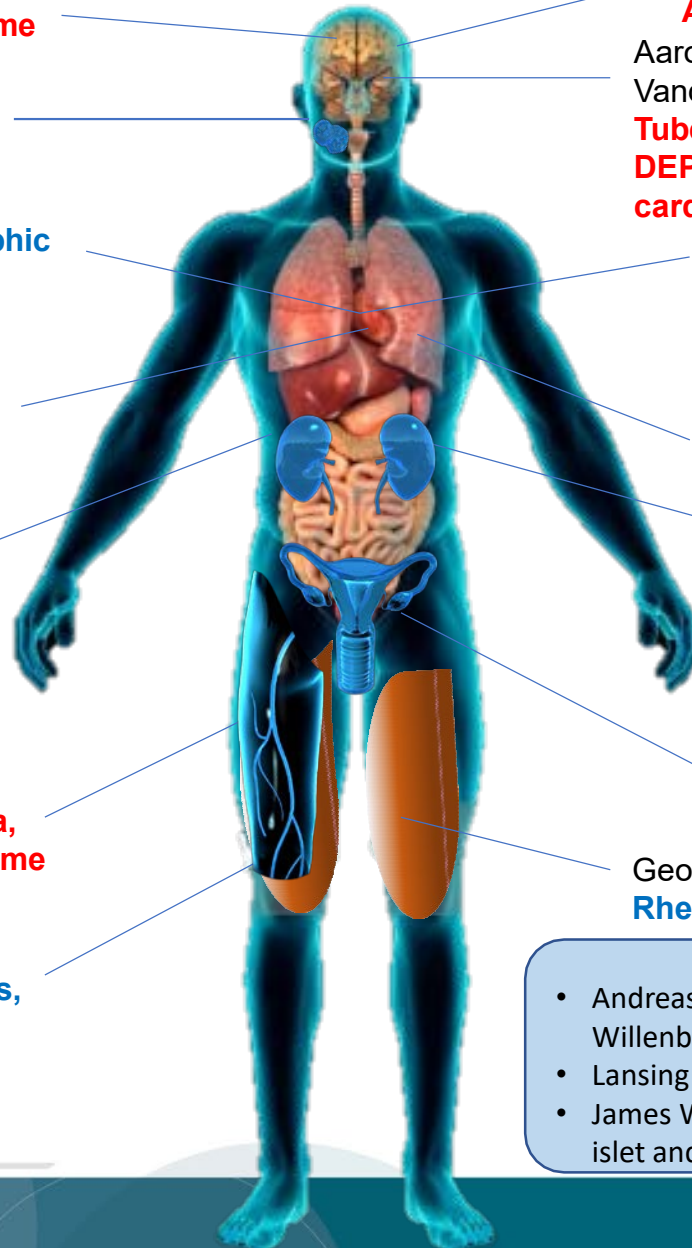
Polycystic ovarian syndrome

George Truskey, Duke U

Rheumatoid arthritis, atherosclerosis

Type-2 Diabetes Mellitus

- Andreas Stahl, Kevin Healy, Matthias Hebrok, Edward Hsiao, Holger Willenbring, UC Berkeley - Pancreatic islet, liver, adipose
- Lansing Taylor, U Pittsburgh – Vascularized liver and pancreatic islets
- James Wells, Moo-Yeal Lee, Cincinnati Children's Hospital - Liver, pancreatic islet and intestine



Advancing understanding of translation:

The Drug Discovery, Development and Deployment Map

- **Problem:** many stakeholders unaware of complexity of developing therapeutic interventions; systems engineers lack common framework for improvements
- **Solution:** create an accurate portrayal of the translational process to educate stakeholders and identify opportunities for innovation
- **NASEM** Forum on Drug Discovery, Development and Translation developed initial map
- Development continuing to incorporate “GoogleMaps-like” features (“traffic”, “GPS”, programs to assist)
- Freely-available at <https://ncats.nih.gov/translation/maps>

NATURE REVIEWS | DRUG DISCOVERY

A dynamic map for learning, communicating, navigating and improving therapeutic development

John Wagner, Andrew M. Dahlem, Lynn D. Hudson, Sharon F. Terry, Russ B. Altman, C. Taylor Gilliland, Christopher DeFeo and Christopher P. Austin

Nat Rev Drug Discov (2018);17:150

CTS Clinical and Translational Science
An Official Journal of ASCPT

ARTICLE

Application of a Dynamic Map for Learning, Communicating, Navigating, and Improving Therapeutic Development

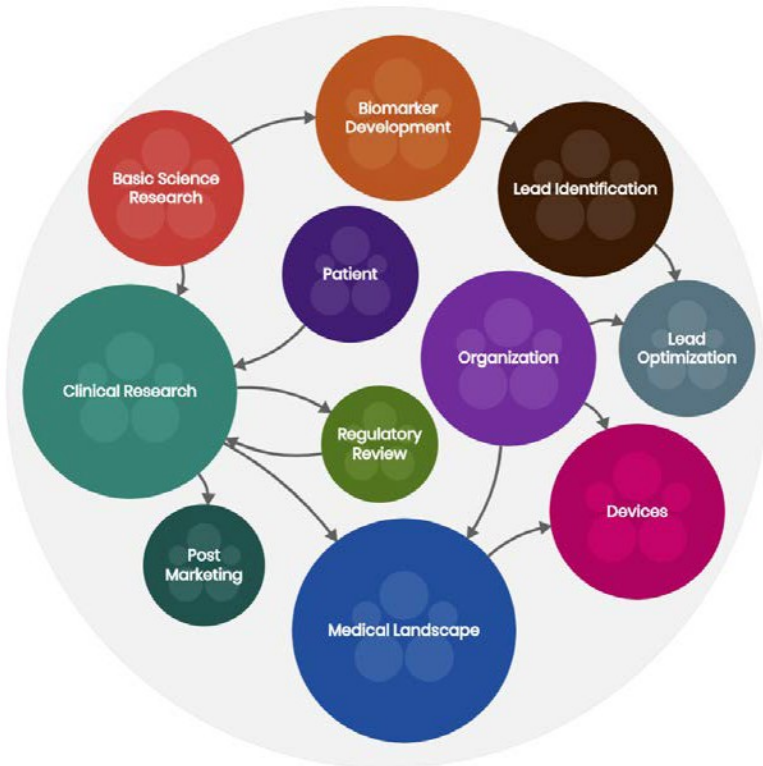
Clin Transl Sci (2018); 11:166



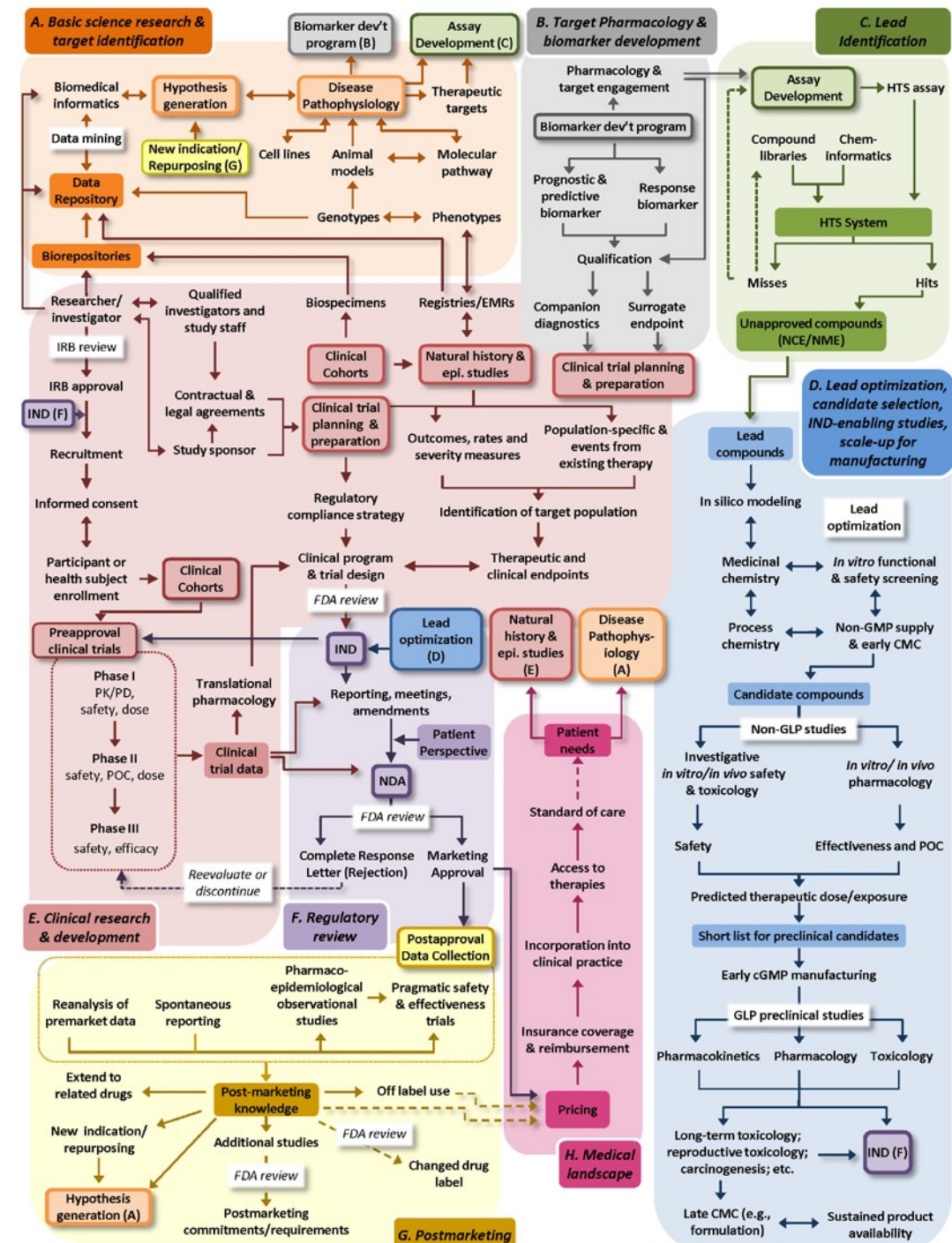
Drug Discovery, Development and Deployment Map



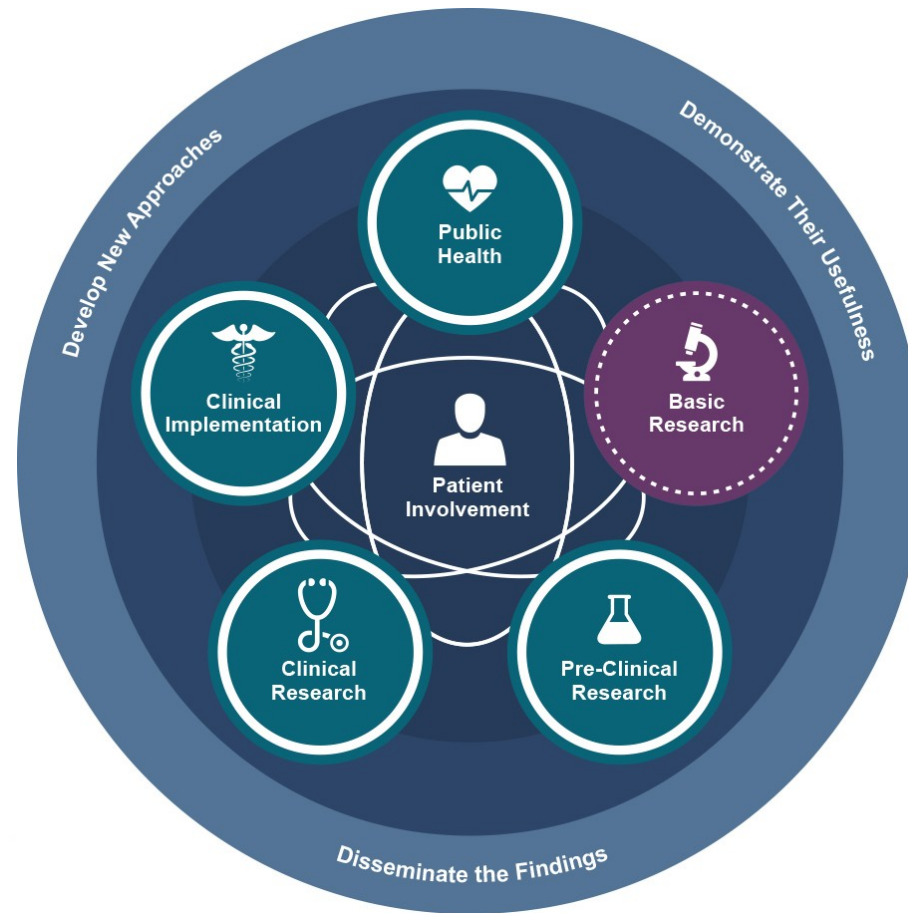
Nature Reviews Drug Discovery 15:150, 2018



<https://ncats.nih.gov/translation/maps>



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