



HYBRID PUBLIC MEETING

Patient Voice in Rare Disease Medical Product Development

October 16, 2026 • 1-5 pm ET

This project is supported by the Food and Drug Administration (FDA) of the U.S. Department of Health and Human Services (HHS) as part of an award of \$135,770 in federal funds (100% of the project). The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by FDA, HHS, or the U.S. Government. For more information, please visit [FDA.gov](https://www.fda.gov).





Welcome

Susan C. Winckler, RPh, Esq.

CEO

Reagan-Udall Foundation for the FDA

WHY WE ARE HERE

Today's meeting centers the patient voice across the development journey

We will examine where patient perspectives are sought, how they shape medical product development, and where additional opportunities for engagement may exist.

The goal is not to prescribe the answer today. It is to surface experience that can inform recommendations for developers and regulators.

BIOLOGICS

DRUGS

DEVICES

ONE CONVERSATION | THREE PRODUCT AREAS

A shared commitment to patient-centered
development

BEFORE WE BEGIN

Housekeeping



Joining Online

Microphone and video will remain off during the meeting

Please share your questions using the Zoom Q&A function



Attending in Person

Please silence mobile devices for the duration of the meeting

Please share your questions using the index cards provided



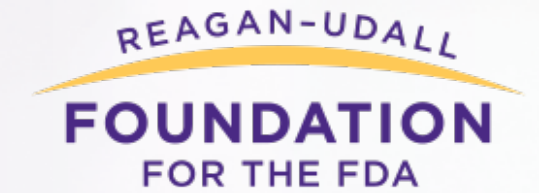
Recording

This public meeting is being recorded

Slides, transcript, and video will be available at www.ReaganUdall.org

Agenda

ALL TIMES EASTERN



1:05pm Opening Remarks

1:10pm CBER & CDER Hub Co-Chairs Discussion

1:30pm **Panel Discussion**
Grounding in Existing Mechanisms: Where the Patient Voice Is Sought Across the Development Timeline

2pm Case Study Presentations

2:40pm Break

2:55pm **Panel Discussion**
Advancing the Patient Voice in Medical Product Development: Reflections and Opportunities

4:45pm CDRH Hub Co-Chair Discussion

4:55pm Closing / Adjourn



Opening Remarks

Amy Comstock Rick, JD

Director, Rare Disease Innovation Hub

FDA

INFOGRAPHIC

DISCUSSION

CBER & CDER HUB Co-chairs



Michael Davis, MD, PhD

Director, Center for Drug Evaluation and
Research (CDER)

FDA



Karim Mikhail, B.Pharm, MSc

Director, Center for Biologics Evaluation and
Research (CBER)

FDA

PANEL DISCUSSION

Grounding in Existing Mechanisms

Where the patient voice is sought across the development timeline



Robyn Bent, MS, RN

Director, Patient-Focused Drug Development
Program

FDA/CDER



Cynthia Grossman, PhD

Director, Division of Patient-Centered
Development

FDA/CDRH



Carmen Matos, MA

Senior Health Scientist, Office of Public
Engagement

FDA/OC/OPA

PRESENTATIONS

Case Studies

Presented in speaking order, followed by moderated Q&A as time allows



Terry Pirovolakis

Founder & CEO

Elpida Therapeutics / CureSPG50



Tracy Gray, MBA, MS, RN

Health Scientist, Patient Engagement Lead

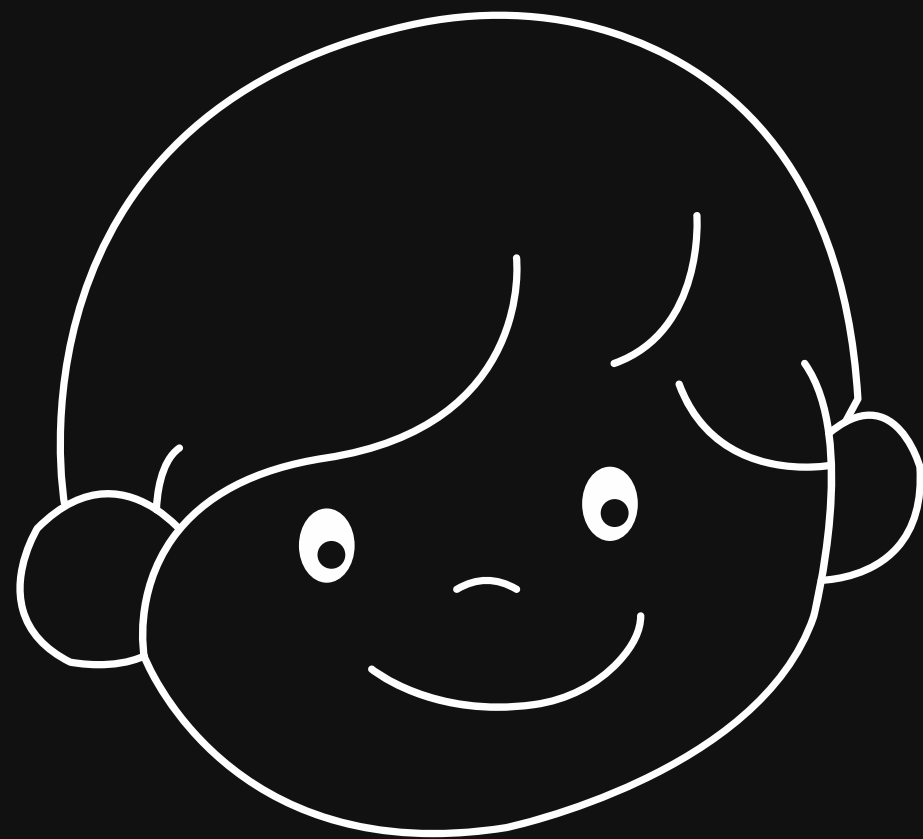
FDA/CDRH



Emily Machiela-Leestma, PhD

Coordinator

HD Benefit-Risk Initiative



#CureSPG50

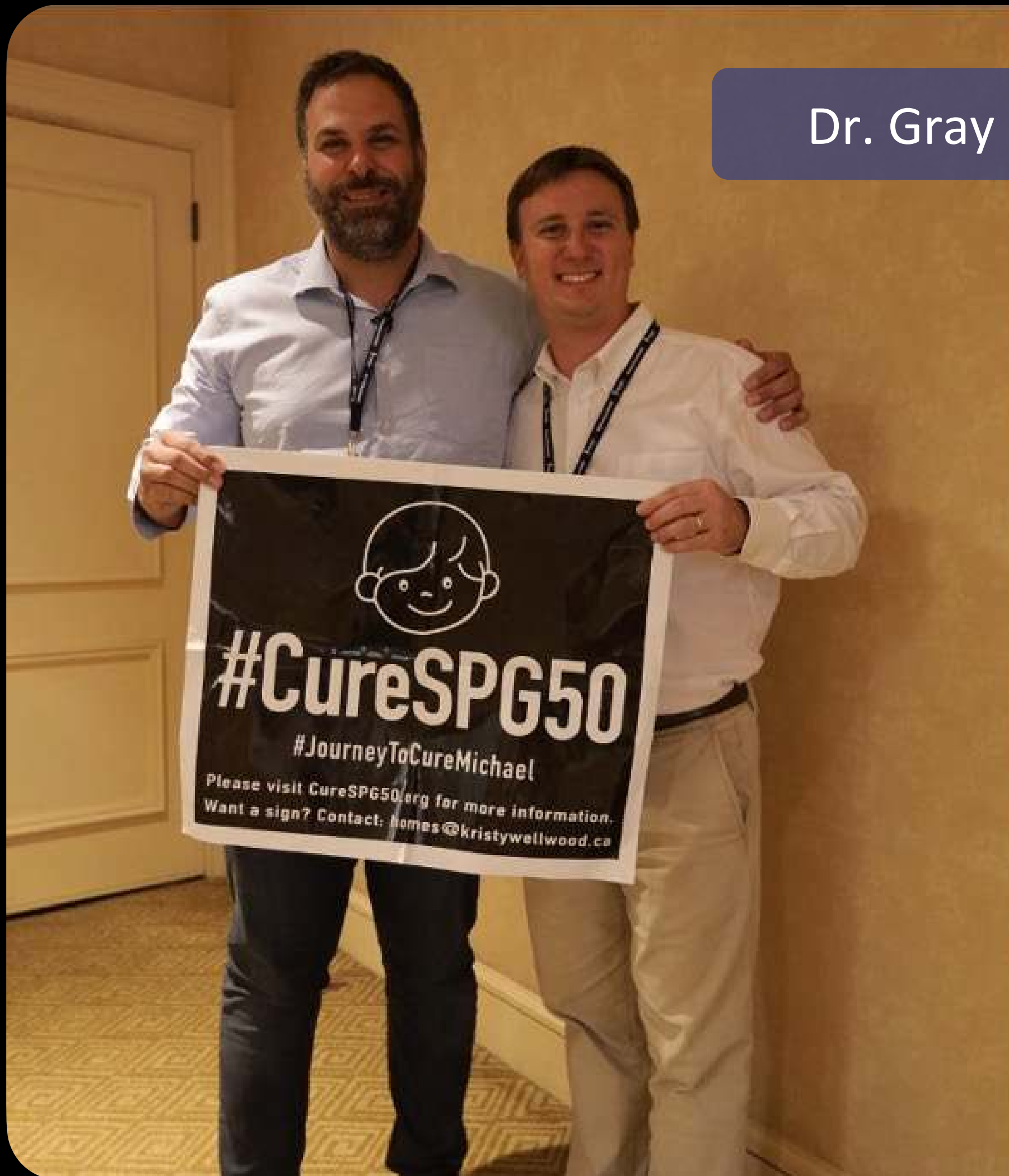
#JourneyToCureMichael



April 2, 2019

Diagnosed with
Spastic Paraplegia Type 50

Dr. Gray



Dr. Chen





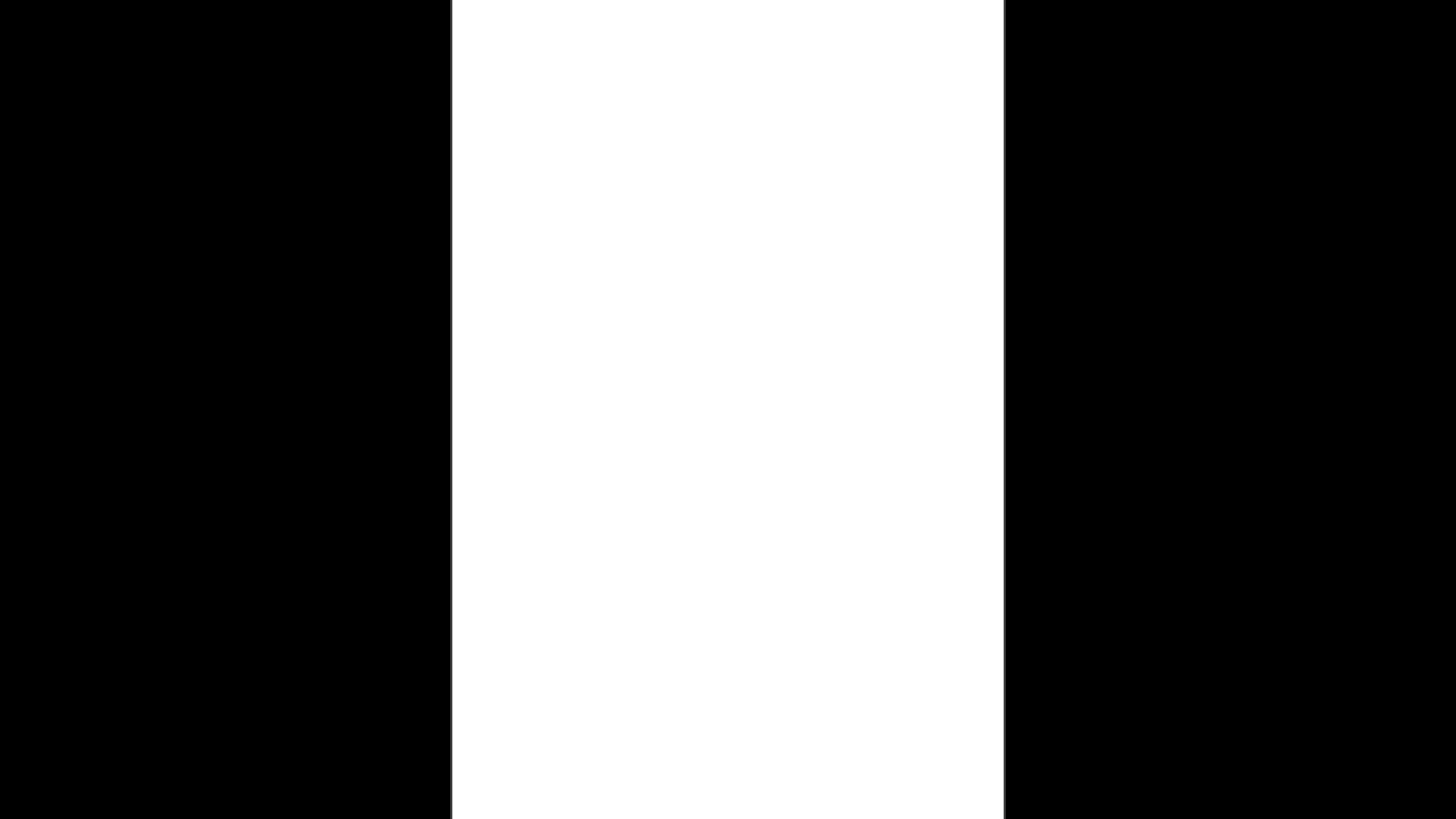
JOURNEY TO CURE MICHAEL FUNDRAISING EVENT

July 28th 3-6pm
Brunswick Bierworks
25 Curity Avenue, East York

Join us for a fun-filled family
afternoon of live music, food,
games and so much more.
Find out more at:
#cureSPG50









Mouse Efficacy Studies

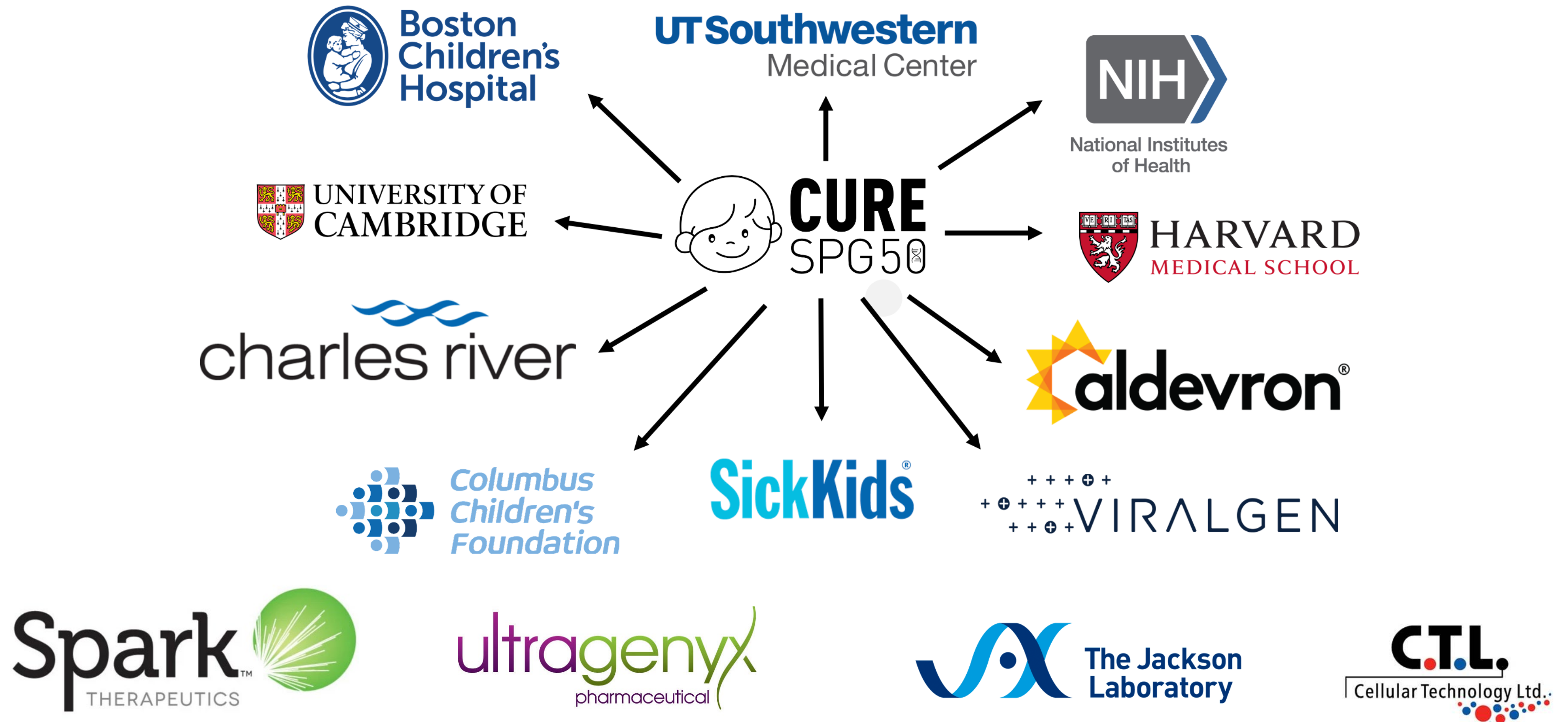


Unaffected



SPG50 Present

Collaborations & Support



Health Canadian CTA Approval



Health
Canada

Health Products
and Food Branch

Santé
Canada

Direction générale des produits
de santé et des aliments

Biologic and Radiopharmaceutical
Drugs Directorate
100 Eglantine Driveway
LCDC Building,
Tunney's Pasture, A.L. 0601C
Ottawa, Ontario
K1A 0K9

Health Canda

No Objection Letter

December 30, 2021

Dec 30, 2021

NO OBJECTION LETTER

Dear Mr. Kenney:

The Clinical Trial Application for Melpida (AAV9/AP4M1)/ Rapamune (sirolimus)/ Prograf (tacrolimus), Control # 259194, concerning Protocol # MELPIDA FOR SPG50, has been reviewed.

In accordance with Part C, Division 5 of the *Food and Drug Regulations*, there is no objection to initiating this trial.

Dosing Day

March 24, 2022



FDA IND Approval

FDA

Study May Proceed

Aug 11, 2022

Diane Balderson

From: [REDACTED]
Sent: Thursday, August 11, 2022 3:18 PM
To: Diane Balderson
Subject: IND 28202 | Study May Proceed
Signed By: [REDACTED]

Dear Dr. Balderson,

We have reviewed your IND 28202 and your study may proceed. This email should satisfy your IRB's requirement for written confirmation from FDA. As a reminder, please be sure to submit all documents that have been exchanged via email during the 30-day review period as an amendment to your IND. If there are any additional non-hold comments, you will receive them in a forthcoming e-mail.

Please acknowledge receipt of this email.

Warm Regards,

[REDACTED]
Regulatory Project Manager (RPM)
Division of Regulatory Project Management
Office of Tissues and Advanced Therapies
Center for Biologics Evaluation Research
U.S. Food and Drug Administration

[OTAT LEARN – Video Instructional Webinars for Sponsors](#)
[Cellular & Gene Therapy Products | FDA](#)

OTAT Mission: The Office of Tissues and Advanced Therapies (OTAT) promotes the public health through collaborative, science-based regulation of medical products. This includes facilitating drug development and ensuring safety of individuals. OTAT's regulatory decisions are data-driven, impartial, and compassionate.

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It Takes A Village





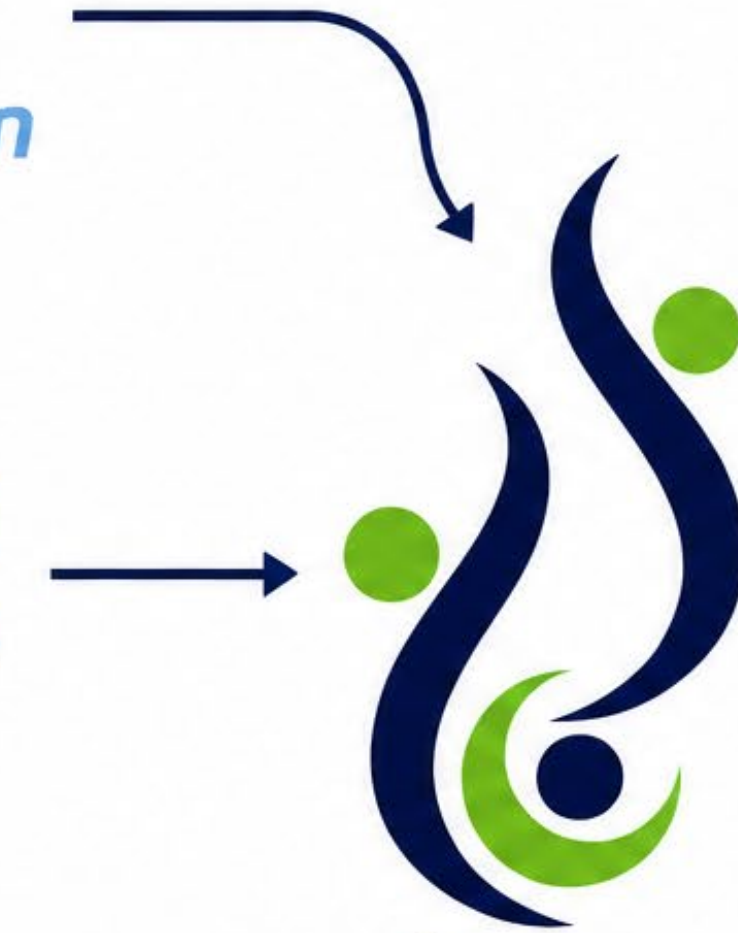
*Columbus
Children's
Foundation*



*fundación
Columbus.*



CURE
SPG50

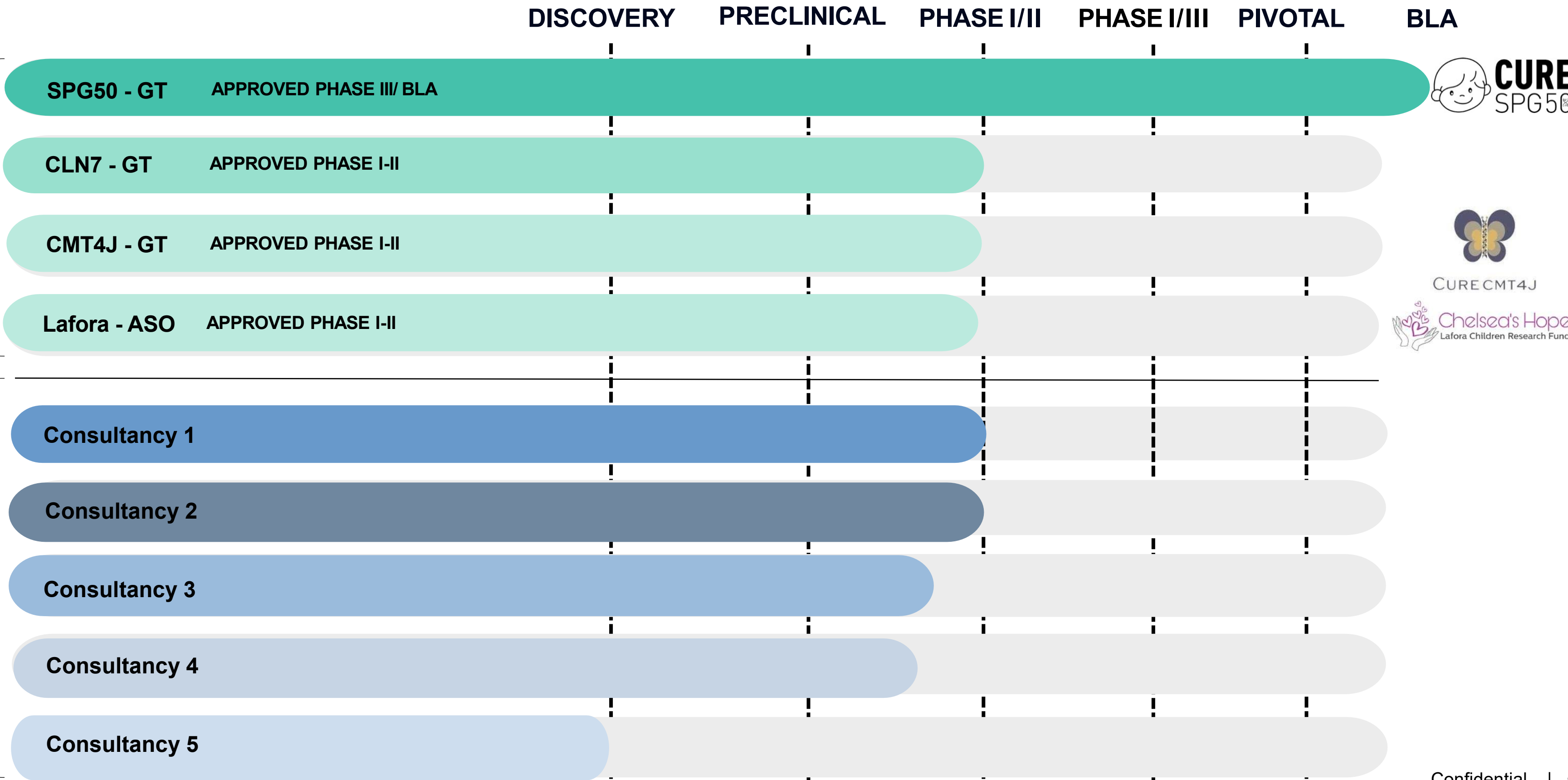


ELPIDA
THERAPEUTICS

Pipeline

Pipeline

Lifeline\Consulting



Partners

Hospitals

UT Southwestern
Medical Center

SickKids[®]

SJD
Sant Joan de Déu
Fundació de Recerca

 **University of Iowa Stead Family Children's Hospital**

NIH
National Institute of Mental Health

 **Azienda Ospedaliera Papa Giovanni XXIII Bergamo**

 **Children's Hospital of Philadelphia**SM

 **Rigshospitalet**

 **Stanford MEDICINE**

 **St. Jude Children's Research Hospital**
Finding cures. Saving children.

 **Boston Children's Hospital**
Until every child is well[™]

 **GREAT ORMOND STREET HOSPITAL CHARITY**

Non-Profits

 **fundación Columbus.**

CIRM
CALIFORNIA'S STEM CELL AGENCY

 **CURE SPG50**

 **Columbus Children's Foundation**

 **eleThon**
COMBATTI LA DISTROFIA MUSCOLARE E LE ALTRE MALATTIE GENETICHE

 **CMT Research Foundation**

 **GENETHON**
CURE THROUGH INNOVATION

 **JACK'S CORNER**

 **CURE CMT4J**
ADVANCING GENE THERAPY FOR RARE DISEASES

CMTA
Charcot-Marie-Tooth Association
ACCELERATING RESEARCH • EMPOWERING PATIENTS

Companies

Catalent[®]

 **Advanthera**
BIOPHARMA SOLUTIONS

 **C. VENTURE PARTNERS**

 **QUATRE** LAB

 **BioSensics**[™]

 **BIOMAPAS**

 **aldevron**[®]

 **charles river**
++ ⊕ ++

ChilliPharm
CLINICAL TRIAL VIDEO SERVICES

VIRALGEN

ultragenyx
pharmaceutical

KO LAW

 **DHC**
DARK HORSE CONSULTING

 **citizen health**

 **jsb**

 **RealTime**
eClinical Solutions

 **KIVO**

 **colpitts**

 **Roche**

TRANSPERFECT

Children Treated

16

SPG50

1

CLN7

1

CMT4J



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50



SPG50

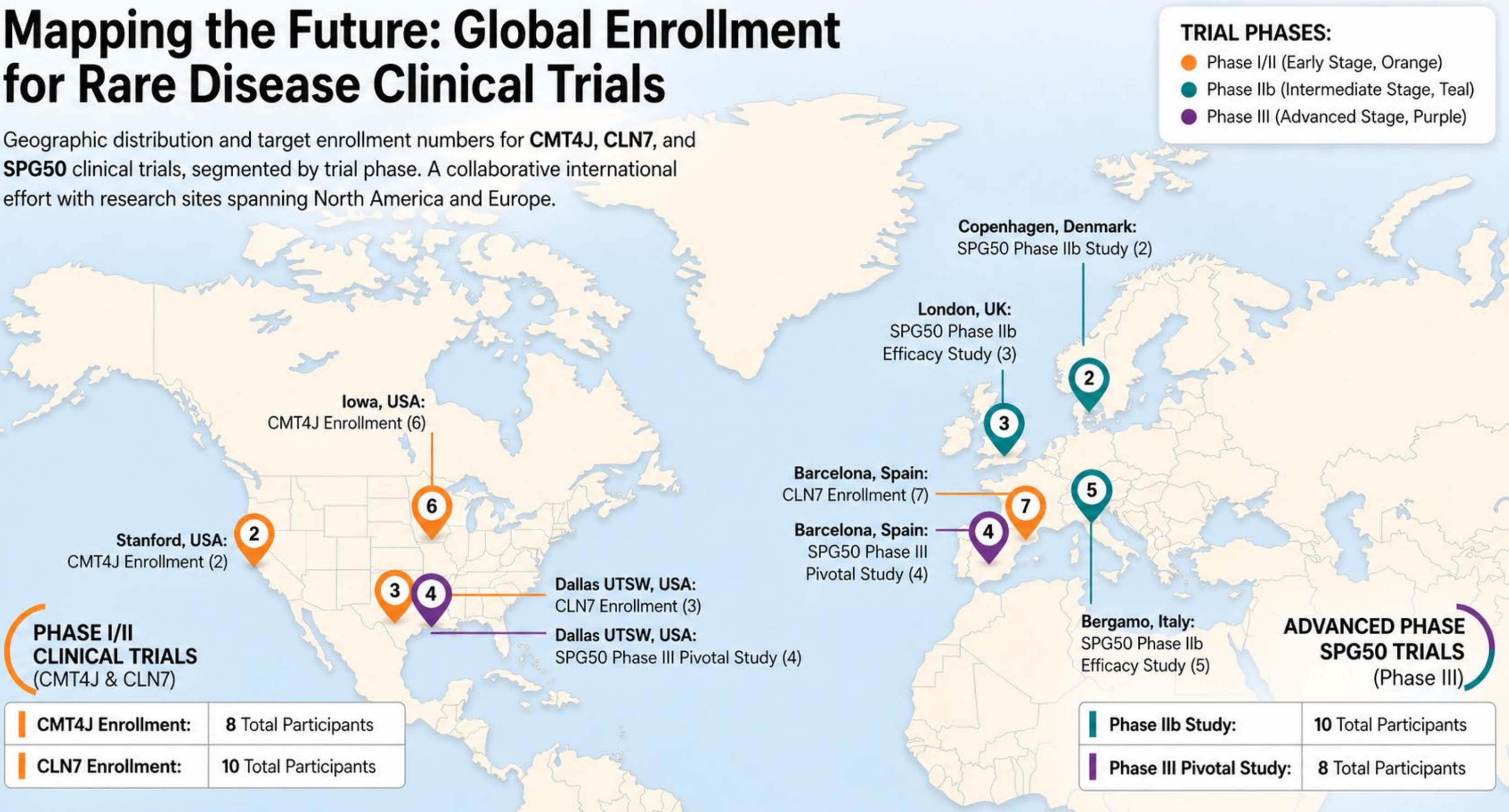


CLN7

Clinical Trial Plan

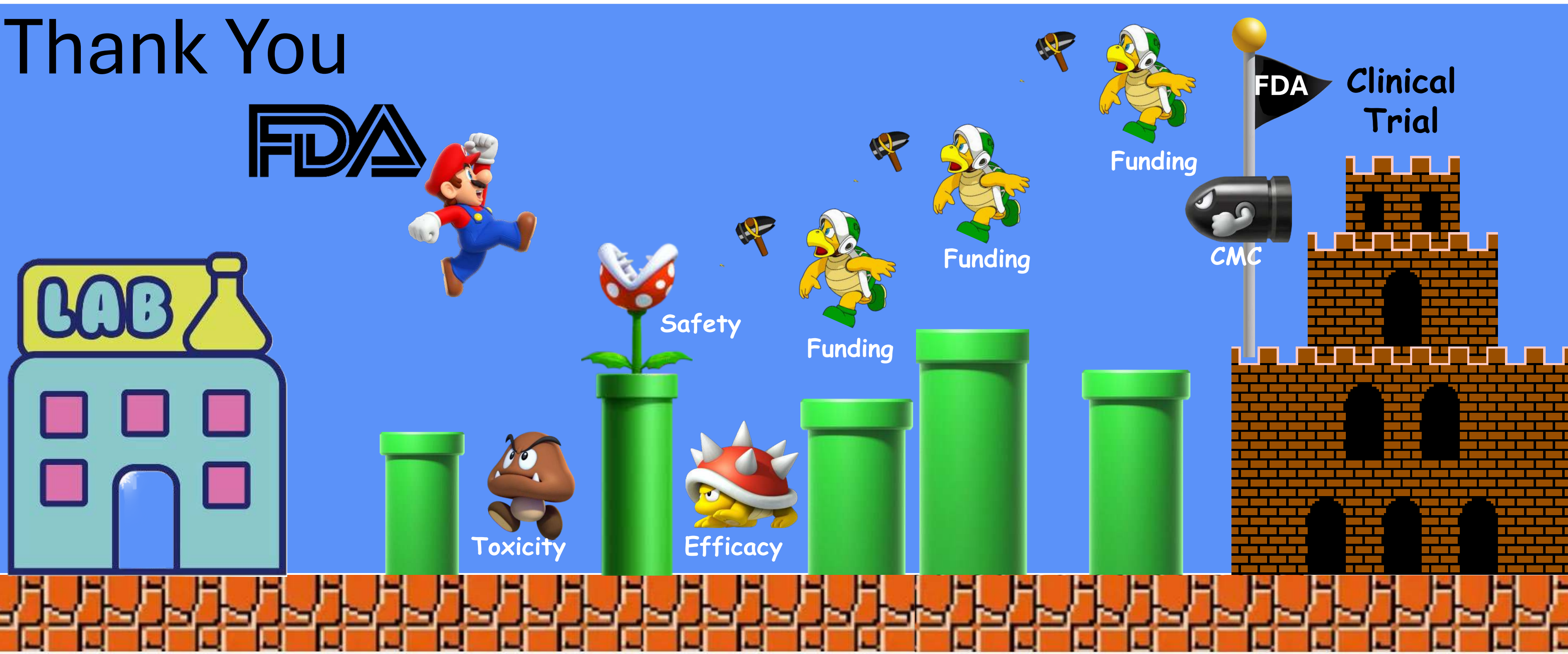
Mapping the Future: Global Enrollment for Rare Disease Clinical Trials

Geographic distribution and target enrollment numbers for **CMT4J**, **CLN7**, and **SPG50** clinical trials, segmented by trial phase. A collaborative international effort with research sites spanning North America and Europe.





Thank You



Drug Approval

Starting
Clinical
Trials



Lessons Learned

- 1 Patient voice from Day 1**
Include PAG in product, trial and regulatory decisions.
- 2 Set realistic therapy expectations**
Define success: improvement, stabilization, slowing progression or prevention.
- 3 Get natural history right (Or the best that can be afforded)**
Poor or incomplete natural history can undermine the entire program.
- 4 Choose meaningful endpoints (Simple & Straightforward)**
The wrong endpoint can make an effective therapy look ineffective.
- 5 Keep listening over time (Phase I-> II->III)**
Patient priorities and benefit-risk tolerance change with disease progression.
- 6 Never Stop Advocating**

Patient engagement should shape the entire product lifecycle—not simply react to it.

FDA

We Need You
NOW!

Ultra-rare should be considered a different regulatory pathway to rare disease. Specific regulations for an ultra-rare path to approval should be created to account for diseases that are not commercially viable to provide a less stringent route to approval (e.g. qualification rather than validation of assays, use of clinical GMP material for commercial sale).

Those that are not commercially viable have no option but to treat under a perpetual IND due to the \$30M+ price to accomplish CMC activities, especially in gene therapy, which is an unsustainable model that ultimately fails both the sponsor and the patients it is meant to serve.

Thank You



CDRH PATIENT ENGAGEMENT USE CASE EXAMPLE

*Tracy Gray, MS RN, MBA
Health Scientist, Patient Engagement Lead
Center For Devices and Radiological Health
Office of Strategic Partnerships and Technology Innovation
Division of Patient-centered Development*

CDRH PATIENT AND CAREGIVER CONNECTION (PCC)

PARTNERS



26 CURRENT PARTNERS

CONTEXT



Growth of home healthcare has resulted in more frequent use of medical devices outside of the professional healthcare facility



Home Use devices are becoming more common and more complex



FDA wants to ensure devices are safe and effective for home or alternative use settings.



HOME USE MEDICAL DEVICES USED BY PANELISTS



Respiratory Devices

Home ventilator
Continuous positive airway pressure
At home pulse-oximeter



Neurological and Mobility Devices

Spinal cord stimulator
Vagus Nerve Stimulator (VNS)
Cane, knee-ankle-foot-orthotic (KAFO)
Ventriculoperitoneal shunts



Gastro devices

Feeding tubes (gastronomy tube,
nasogastric (NG) feeding tube)
Total parenteral nutrition (TPN) infusion
Insulin pump used to deliver cortisol
Ostomies (ileostomy, jejunostomy)



Diagnostic devices

COVID-19 at home testing



Other

Wound vac for fistulas
Holter monitor machine



CDRH Patient Engagement Town Hall

PERSPECTIVES ON HOME USE
DEVICES BY THE NATIONAL
ORGANIZATION OF RARE DISEASE
(NORD) COMMUNITY

Wednesday, September 13, 2023

PATIENT PANEL KEY FINDINGS

The FDA logo, consisting of the letters "FDA" in white on a blue square background.

**FACILITATE BETTER
QUALITY OF LIFE**

**FLEXIBILITY ENABLING
PARTICIPATION IN
ACTIVITIES**

**CONCERNS ABOUT
TRANSITIONING FROM
TRADITIONAL
HEALTHCARE SETTING**

**INFORMATION FOR OFF-
LABEL USE**

**ALIGNMENT OF DEVICE
FUNCTION WITH LIFESTYLE**

**UPDATED INFORMATION
AS CONDITION
PROGRESSES**

**DEVICES THAT MEET
HEALTH EQUITY NEEDS**



THANK YOU!

Emily Machiela-Leestma, PhD

Coordinator

HD Benefit-Risk Initiative



BREAK

**We'll resume at
2:55pm ET**

Please return promptly

PANEL DISCUSSION

Advancing the Patient Voice

Reflections and opportunities



Allyson Berent, DVM
Chief Science Officer
Foundation for Angelman Syndrome Therapeutics



Michelle Campbell, MS, PhD
Associate Director for Stakeholder Engagement & Clinical Outcomes
Office of Neuroscience, OND/CDER/FDA



Donald "DJ" Carlino
Manager, Strategic Alliances
Novocure



Tom Croce, BSPHarm
Vice President, Global Patient Advocacy & Engagement
Jazz Pharmaceuticals



Mona Elmacken, MD
Lead Physician, Malignant Hematology Branch
OTP/CBER/FDA



Courtney Lias, PhD
Director, Office of In Vitro Diagnostics
OPEQ/CDRH/FDA



Jenn McNary
Principal & Co-Founder
Canary Advisors



Tamar Thompson, MS
Vice President, Patient Experience & Insights
Alexion, AstraZeneca Rare Disease



Ashley Valentine, MRes
Co-founder & CEO
Sick Cells



Final Discussion & Closing

Michelle Tarver, MD, PhD

CDRH Director

FDA

Thank you for advancing patient-centered development

This project is supported by the Food and Drug Administration (FDA) of the U.S. Department of Health and Human Services (HHS) as part of an award of \$135,770 in federal funds (100% of the project). The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by FDA, HHS, or the U.S. Government. For more information, please visit [FDA.gov](https://www.fda.gov).