HEREDITARY NEUROPATHY FOUNDATION

cmtupdate

Impact That Matters

Driving CMT Research with the Patient's Voice

SPRING 2024

CMT Clinical Trial Readiness Summit

Pg 3

CMT UPDATE
SPRING 2024

Dear HNF Supporter,

A lot is going on at HNF! We have a new member of our Board of Directors to introduce, and we are ramping up fundraising activities to help expand on the impactful work that the HNF does for our CMT community.

First, here's a message from our newest member of the Board of Directors, Dennis Sullivan. →

Next, to substain the impactful initiatives undertaken by HNF, generous contributions from supporters like you are crucial. Your support isn't just greatly appreciated; it's essential for enabling HNF to continue its remarkable and patient-centric research.

HERE, you'll find a comprehensive review detailing all of HNF's accomplishments throughout 2023, along with our strategic focus for the year ahead in 2024. We trust you'll find this update informative and compelling, reinforcing our collective commitment to advancing HNF's mission.

We extend our heartfelt gratitude for your steadfast support, both past and present. With your continued dedication, let's strive to make 2024 the most remarkable year in HNF's history yet. Thank you for being an invaluable part of our journey towards making a difference.

Warm regards,

Allison T. Moore

Allison T. Moore

Hello CMT Family,

I am writing to introduce myself to you. My name is Dennis Sullivan, and I am the newest member of the Board of Directors at HNF. I became involved with this impactful organization last year while working on a presentation about CMT that I was giving to an employee resource group at my company, Medtronic, Inc. I contacted HNF for help and Estela Lugo and Courtney Hollett supported me every step of the way, even participating in giving the presentation.

The next steps came naturally. I have long been looking for a way to give back to my community for the good fortune I have had in my life, so I asked HNF what I could do to support their efforts. With CMT in my family and a 35-plus year career in the medical device industry, primarily in orthopedics and spine, and most recently in surgical robotics and AI, HNF was a natural fit for my goal.

Fast forward six months, and here we are. As a new Board Member with a passion to help HNF advance its mission. It is truly an exciting opportunity, and I am already jumping in with both feet. No task is too small and none too daunting. There is a lot to do, but I am ready for the challenge.

Warmest regards,

Dennis Sullivan

HEREDITARY NEUROPATHY FOUNDATION

HNF Accomplishments:
Click Here!

Donate to HNF:
Click Here!

HNF Announces: CLINICAL TRIAL READINESS SUMMIT

HNF is thrilled to announce its first CMT Clinical Trial Readiness Summit to be held June 7-8 in San Diego, CA. As new clinical trials emerge for CMT, optimizing outcome measures, endpoints, and the participant experience has never been more urgent! HNF is committed to derisking upcoming trials by bridging the gap between clinical trial study teams and patient perspectives and experiences.

As HNF focuses on accelerating access to critical research data and specimens, this summit will include on-site CMT Biobank collection and a digital wearable pilot study sponsored by BioSensics™ and HNF using FDA-registered wearable devices. HNF will conduct objective assessments of gait, balance, fall risk, mobility, and upper limb function.

Attendees will also have opportunities for 1-on-1 consultations with CMT experts like Glenn Pfeffer, MD, on surgery and Mitchell Warner, CPO, on Helios Bracing. InformedDNA is providing discounted telehealth visits for genetic counseling and access to patient-initiated genetic counseling during this event. CMT Lounges will be open across both days for building friendships and connecting with CMT resources like genetic testing, adaptive exercise, mental health, the GRIN patient registry, and more.

This impact-driven summit is a chance for all patients, caregivers, clinicians, industry, and regulatory representatives to connect with one another, exchange ideas, and engage in open discussions about the obstacles of designing and conducting successful CMT clinical trials.

The summit will be an all-in-one event for CMT Clinical Trial readiness with patients at the heart and center!

Attendees can expect to:

- Have the chance to participate in a pilot study sponsored by BioSensics™ and HNF using FDA-registered wearable devices for objectively assessing gait, balance, fall risk, mobility and upper limb function.
- Share their lived experience and patient/caregiver perspective during clinical trial design brainstorming sessions with industry partners currently planning human trials.
- Donate blood/urine samples to the CMT Biobank for expedited access to researchers currently working on CMT treatments.
- Connect and socialize with other individuals living with CMT.
- Learn what it takes to be part of a CMT clinical trial and the latest CMT research.

Speakers include:

Shoshana Shendelman, PhD, CEO/ Founder, Applied Therapeutics

Klaus Romero, MD, MS, FCP, CEO, Critical Path Institute

Kayla Cornett, PhD, University of Sydney

Ashkan Vaziri, PhD, Founder/CEO, BioSensics™

Allison Moore, Founder/CEO, Hereditary Neuropathy Foundation

Glenn Pfeffer, MD, Cedars-Sinai Orthopaedic Center

Matthew Jarpe, PhD, Scientific Advisor, Hereditary Neuropathy Foundation

Anna Pfalzer, PhD, CSO, COMBINEDBrain

Lori Sames, Founder, Founder/CEO, Hannah's Hope Fund

Joy Aldrich, GRIN Registry Coordinator, Hereditary Neuropathy Foundation

Robert Moore, GRIN Data Manager, Hereditary Neuropathy Foundation

Jason Colquitt, CEO, AcrossHealth Care

Gideon Shapiro, PhD, Founder/CSO, Miralinc Pharma

Shannon Strom, PhD, RAC, Drug Development Expert

Michelle Campbell, PhD FDA, Associate Director, Stakeholder Engagement & Clinical Outcomes, Office of Neuroscience

Tim Estilow, OTR/L, CHOP

Dottie Caplan, SVP Patient Advocacy and Engagement, Applied Therapeutics

Lori Sames, Founder, Hannah's Hope Fund

Sharon Hesterlee, PhD, Chief Research Officer, MDA

Sabrina Yum, MD, CHOP

Brian Lin, PhD Director, Research Portfolio, MDA

Estela Lugo, Program Development Manager, HNF

Julie Stone, All Bodies Community

Constantine Farmakidi MD, University of Kansas Medical Center

Session Titles include:

Putting CMT Blood Samples to Work

For Industry & Researchers Only: Surrogate Biomarkers

For Patients & Caregivers: Workshop Managing Mental Health & CMT

Measuring CMT with Wearable Technology

Revolutionizing CMT Clinical Trials with Video

Is CMT Surgery Right for You?

Identifying Roadblocks to CMT Clinical Trials

Turning Symptoms into Science

What Does it Take? A Fighter Mom's Journey from Kitchen Table to Injection!

What's Next for Gene Therapies in CMT?

Overview: Upcoming CMT Pipeline & Clinical Trials

Philanthropic & Investment Opportunities

For more information and to register: CLICK HERE!

A special thanks to HNF's Summit Sponsors:

Applied Therapeutics, NMD Pharma, Novartis, Alesta Therapeutics, BioSensics™, Miralinc Pharmaceuticals, Matrix, EveryLife Foundation For Rare Diseases, Helios Bracing, and COMBINEDBrain.

Summit sponsorships are still available. Please email Courtney Hollett at courtney@hnf-cure.org to learn more.

2024 CMT Clinical Trial Readiness Summit
DAY 1 AGENDA FRIDAY, JUNE 7, 2024

8:00 AM: REGISTRATION & BREAKFAST

9:00 – 9:20 AM: Keynote Address
From NEW Gene to Phase 3 Clinical Trial – Treatment is Within Reach!
Shoshana Shendelman, PhD
CEO & Founder, Applied Therapeutics

9:30 – 10:45 AM: Panel Discussion
CMT Biobank & Biomarkers for CMT Therapy Development
A biomarker may be used to predict which patients will respond to treatment or see how well the body responds to a treatment for a disease. Without them, clinical trials may enroll patients who will have a lower chance to respond, or they rely on clinical evaluations that may not be sensitive enough to capture change (good or bad). In this session, you'll hear from experts in the field on the importance of identifying and validating biomarkers for use in CMT therapeutic development. What's on the horizon, and how is a patient-led biorepository advancing the development of evidence-based biomarkers for all CMT subtypes?

MODERATOR: Matthew Jarpe, PhD, HNF Scientific Advisor
Anna Pfalzer, PhD, CSO, COMBINEDBrain
FIRESIDE CHAT PANELIST: Anna Pfalzer, PhD, CSO, COMBINEDBrain
Shoshana Shendelman, PhD, CEO & Founder, Applied Therapeutics
Allison Moore, Founder/CEO, Hereditary Neuropathy Foundation

Roundtable Discussions & Feedback Sharing

10:45 – 11:00 AM: Break

11:00 – 12:15 PM: Panel Discussion
Innovating Digital Endpoints with Wearable Technology Development
The identification and development of digital measures are vital to remote monitoring CMT disease progression or improvement in real-world settings. By collecting remote data (not in a clinical setting), researchers, industry, and regulatory agencies can better understand the daily impact of CMT on people living with the disease. This panel session will cover the development of methods to identify digital measures of CMT disease from wearables to generate clinically meaningful patient outcomes. Participants will have the option to participate in a digital wearable pilot study sponsored by BioSensics and HNF.

MODERATOR: Klaus Romero, MD, MS, FCP, CEO, Critical Path Institute
Kayla Cornett, PhD, University of Sydney
Ashkan Vaziri, PhD, Founder and CEO, BioSensics™
CMT Patient, TBD

Audience Participation Q & A

12:15 – 1:00 PM: LUNCH

1:00 – 2:15 PM: Panel Discussion
Revolutionizing Clinical Trials with Video – If a Picture is Worth a Thousand Words, a Video is Worth a Million
To make clinical trials for CMT successful, we need new ways to measure how the disease affects people. One idea is to use videos to see how it affects things like walking and step length. Using videos could allow us to use footage from the past or even videos taken by regular people, not just clinic experts. Given the increased use of telemedicine, video may be a useful addition to clinical trial assessments.

MODERATOR: Klaus Romero, MD, MS, FCP, CEO, Critical Path Institute
Allison Moore, Founder/CEO, Hereditary Neuropathy Foundation
PT, DPT, CMT Centers of Excellence (pending approval)
Michelle Campbell, PhD FDA, Associate Director, Stakeholder Engagement & Clinical Outcomes, Office of Neuroscience

Patient Video Breakout Session & Feedback Sharing

2:15 – 2:45 PM: CMT Lounges:
In-Person Support, Vendor Resources & Socializing!
CMT Social – Meet & Greet w/ Vendor Tables
CMT Adaptive Exercise – **Julie Stone**, Founder, All Bodies Community

2:45 – 3:15 PM: Presentation:
Is CMT Surgery Right for You?
Glenn Pfeffer, MD
Join us for an enlightening summit session titled "Is Surgery Right for You" with renowned orthopedic surgeon Glenn Pfeffer, MD. As Director of the Foot and Ankle Center at Cedars-Sinai Medical Center in Los Angeles, Dr. Pfeffer brings over 30 years of experience in treating foot and ankle problems in patients with CMT and has performed over 1000 CMT foot surgeries.

3:20 – 4:45 PM: Panel Discussion:
Identifying Roadblocks to Success for CMT Clinical Trials
This interactive panel will shed light on the unique challenges faced by CMT clinical trials, including the diverse subtypes & symptoms, limited clinical experience, patient access, investment, and complementary outcome measures that are sensitive enough to measure change in a relatively short period of time. You'll learn about evidence-based, innovative strategies and solutions that may apply to CMT for enhanced efficacy and the future success of CMT clinical trials. The open format will allow for patient engagement interaction to better define how these potential solutions, such as adaptive and decentralized trials, including telemedicine, patients & caregiver needs, real-world evidence (RWE) data capture (patient registries, collaborative networks & wearable & tech devices) may have an impact in supporting the success of these trials.
MODERATOR: Shannon Strom, PhD, RAC, Drug Development Expert
Michelle Campbell, PhD FDA, Associate Director, Stakeholder Engagement Clinical Outcomes, Office of Neuroscience
Sabrina Yum, MD, CHOP
Dottie Caplan, SVP Patient Advocacy and Engagement, Applied Therapeutics (Panelist)
CMT Patient, TBD

Audience Participation Q&A

4:45 – 5:00 PM: Closing Remarks

2024 CMT Clinical Trial Readiness Summit
DAY 2 AGENDA SATURDAY, JUNE 8, 2024

8:00 AM: CHECK IN & BREAKFAST

9:00 – 10:00 AM

Making Sense & Science of CMT Symptoms

"Is this symptom related to CMT?" This is a frequent question asked by many individuals living with CMT and their loved ones. Most of us are familiar with the most common symptoms of CMT, such as drop foot & hand weakness. However, many less-recognized symptoms can be just as disruptive to our lives but have not yet been studied or linked to CMT. This is why in 2013, HNF started the Global Registry for Inherited Neuropathies (GRIN) and the CMT Natural History Study.

How do we know if our symptoms are CMT-related, and how can we drive research to answer this question better?

Joy Aldrich, Grin Registry Coordinator, HNF (Presenter & Panelist)
Jason Colquitt, CEO, AcrossHealthcare
Robert Moore, GRIN Data Manager, HNF

Roundtable Discussions & Feedback Sharing

10:00 - 10:25 AM: CMT Lounges:

In-Person Support, Vendor Resources & Socializing!

CMT Social – Meet & Greet w/ Vendor Tables
CMT Adaptive Exercise – **Julie Stone**, All Bodies Community

10:30 - 10:50 AM: Presentation:

What Does it Take? A Fighter Mom's Journey from Kitchen Table to Injection!

Lori Sames, Founder, Hannah's Hope Fund

Learn how "fighter mom" Lori Sames raised the necessary funds and curated the qualified experts to tackle a gene therapy for Giant Axonal Neuropathy (GAN), an inherited neuropathy. Topics covered will include lessons learned, accelerating progress, cost-effective strategies, and regulatory considerations. Hear from Lori about the next steps needed to cross the finish line and how this will transform the future of gene therapies in CMT.

11:00 - 12:25 PM: Panel Discussion

What's Next for Gene Therapies in CMT?

How does the drug development process work and how long does it take for a drug to get from a research lab to your pharmacy? What is gene therapy and why is it important to develop treatments for specific CMT types? What are the pros and cons of gene therapy? What is currently in the gene therapy pipeline for CMT?

You'll learn the good, the bad and the ugly of gene therapy during this overview of the various approaches. Topics will include targets and delivery options and which subtypes have the best chance of success, Conventional therapeutic approaches vs. gene therapy, can patients participate in both? Other questions answered during this session will include: How does the FDA view gene therapies for CMT? What are the pros & cons, what are the obstacles, and is it right for you? Patients and stakeholders will walk away with the basic knowledge & understanding of what it takes to develop a gene therapy for CMT.

Sharon Hesterlee, PhD, Chief Research Officer, MDA
Sabrina Yum, MD, CHOP
Brian Lin, PhD Director, Research Portfolio, MDA
Matt Jarpe, PhD, Science Advisor, HNF
Lori Sames, Founder, Hannah's Hope Fund

Audience Participation Q & A

12:30 – 1:15 PM: LUNCH

1:15 – 2:15 PM:

For Industry & Researchers Only: Surrogate Biomarkers

Matthew Jarpe, PhD, HNF Scientific Advisor
Anna Pfalzer, PhD, CSO, COMBINEDBrain

For Patients & Caregivers: Workshop Managing Mental Health & CMT

Estela Lugo, Program Development Manager, HNF
Julie Stone, Founder, All Bodies Community

2:30 – 4:00 PM: Presentation:

Overview: Upcoming CMT Pipeline & Clinical Trials

Hear from HNF's Therapeutic Research in Accelerated Discovery (TRIAD) partners about their latest research milestones and what's next in the pipeline for CMT.

Matthew Jarpe, PhD, HNF Scientific Advisor
Gideon Shapiro, PhD, Founder/CSO, Miralinc Pharma - HDAC6 to treat CMT
TBD: pending approvals
TBD: pending approvals

4:00 – 4:30 PM: Philanthropic & Investment Opportunities

4:30 PM: Closing Remarks

5:30 PM: HNF Cocktail & Dinner Party

HEREDITARY NEUROPATHY FOUNDATION

June 7-8, 2024

SAN DIEGO, CA

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HNF Patient-Centered

CMT Clinical Trial Readiness Summit

Reserve your seat today!

CMT Patient Voices

Loews Coronado Bay Resort

The time for CMT Clinical Trials is NOW!

- **Share** your patient perspective for upcoming CMT clinical trials
- **Donate** specimens to the CMT BioBank
- **Learn** what it takes to be part of a CMT clinical trial
- **Connect** with others living with CMT

hnf-cure.org

4000 Coronado Bay Rd, Coronado, CA 92118

BREAKING NEWS

HNF to Host:

"Revolutionizing Clinical Trials" Panel at Bio 2024

HNF is thrilled to announce that we have been accepted to host a 1-hour panel discussion during the Bio International Convention (https://convention.bio.org**) in San Diego, CA, in June, 2024.**

The session titled, ***"Revolutionizing Clinical Trials: Patient Registries, Wearable Tech, and Video Capture"***, will include experts from the FDA, Stanford University, Across Healthcare, and HNF's Founder and CEO, Allison Moore.

The Bio International Convention is the industry's premier event, hosting thousands from across the global biotech ecosystem to focus on the future of bringing treatments to market. HNF's session attendees will gain insight into the dynamic fusion of patient-led registries like GRIN, wearable tech, and video capture, unveiling unprecedented opportunities in clinical trials championed by patient advocates and fostering patient-centric innovation.

This dynamic panel will include::

Moderator:
Klaus Romero, MD, MS, FCP, CEO, Critical Path Institute

Speaker:
Michelle Campbell, PhD, Associate Director, Stakeholder Engagement & Clinical Outcomes, Office of Neuroscience, FDA

Speaker:
Jason Colquitt, CEO,
Across Healthcare

Speaker:
Tina Duong, PhD, Director,
Clinical Outcomes R&D, Stanford University

Speaker:
Allison Moore, Founder/CEO,
Hereditary Neuropathy Foundation

We are excited to be able to showcase this important topic on the "big stage" and represent the Charcot-Marie-Tooth community.

BREAKING NEWS

Where breakthroughs begin

HNF-HOSTED BIO PANEL SESSION

Date
June 3-6, 2024

San Diego
California

Revolutionizing Clinical Trials:
Patient Registries, Wearable Tech, & Video Capture

SPEAKERS:

Allison Moore
HNF

Michelle Campbell, PhD
FDA

Tina Duong, PhD
Stanford University

Klaus Romero, MD, MS, FCP, CEO,
Critical Path Institute

Jason Colquitt
CEO, Across Healthcare

CMT IMPACT

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2024 THE ROAD TO CMT THERAPIES

The road to **CMT therapies** has never been clearer & more achievable than today!

HNF is impacting each stop along the way with crucial programs such as, **TRIAD, GRIN, the CMT Genie & CMT Biobank!**

View our **CMT Road map** to learn more!

Impact that matters.

THE ROAD TO CMT THERAPIES

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Drug/Gene Therapy Development

HNF works with our **TRIAD** partners including innovative biotech, pharma and award-winning researchers to fund and support the most promising scientific approaches to potential treatments.

01 WATCH VIDEO

Natural History Studies

GRIN's Natural History Studies are critical in identifying more patients to understand impact of disease and to support clinical trial design and patient recruitment. The **CMT Genie** supports patients to confirm the genetic mutation of their CMT.

02 WATCH VIDEO

Test in Models

HNF develops and validates CMT cell and animal models to test the efficacy of viable treatment approaches. This includes toxicology and dosing studies. Our **CMT Biobank** collects patients' biospecimens to accelerate crucial biomarker development.

03 WATCH VIDEO

FDA Regulatory

HNF engages with FDA regulators to incorporate innovative and digital technologies to enhance the precision of sensitive data capture. HNF pilot studies serve to accelerate, develop, and validate CMT functional outcome measures for clinical trials.

04 WATCH VIDEO

hnf-cure.org Watch videos: www.hnf-cure.org/our-impact/clinical-trials

HEREDITARY NEUROPATHY FOUNDATION

HNF IMPACT IN 2023 A REVIEW

Human Trials

We thank you from the bottom of our hearts for your support in 2023. Your donations continue to fuel our efforts toward our ultimate finish line: successful human clinical trials for CMT.

DONATE

TRIAD

- 2 Rat Models (GDAP1 & SORD)
- 3 Cell Models (GDAP1, MFN2, MTRFR)
- Repurposed Drug Screens (*See below)
- 3 Novel Drug Milestones Met (MFN2)
- 6 Publications

GRIN

- 5000+ GRIN Participants
- 41 CMT Mutations
- 60 Countries
- 1 Pediatric Natural History Study
- 1 CMT Pain Management Guide

Genie

- 300+ Participants
- 200+ Genetically Tested
- 130+ Diagnosed
- 1 New Virtual Genetic Counseling Partner

Biobank

- 5 Cities
- 65+ Biospecimens Collected
- 3 Exploratory Biomarker Studies

*CMT1A, HNPP, CMT2A, CMT2K, Sord Deficiency, CMT4A, CMT4B, CMT4F, CMT4J, CMT6

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Join Team CMT!

MAKE AN IMPACT WITH US!

HOW TO CONTRIBUTE

- Make a single or a recurring donation.
- Ask your company if they have payroll deductions for charitable contributions and if they match your contributions. (It should be pre-tax, which reduces your taxable income!)
- Participate or host a fundraiser
- Create a Facebook or Birthday Fundraiser
- Have any other ideas? Email us at info@hnf-cure.org

Did you know that HNF has 4 out of 4 stars from Charity Navigator, the largest and oldest charity evaluator?

★★★★ CHARITY NAVIGATOR Four Star Charity

DONATE

BBB

95% of donations Fund CMT Research & Programs

hnf-cure.org

HNF Contributes CMTRN Data to C-Path's RDCA-DAP, Strengthening Research Capabilities for Inherited Neuropathies

Integration of CMTRN data into RDCA-DAP promises new insights into Charcot-Marie-Tooth disease and related neuropathies, fueling advancements in treatment and care.

Recently, C-Path (Critical Path Institute) proudly announced a significant data sharing agreement with the Hereditary Neuropathy Foundation (HNF), integrating valuable data from the Charcot-Marie-Tooth Research Network (CMTRN) into C-Path's Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®). This integration, facilitated through the National Organization for Rare Disorders' (NORD) IAMRARE® registry, represents a significant leap forward in the research and understanding for the ultra-rare forms of Charcot-Marie-Tooth (CMT) disease and related inherited neuropathies.

"There are over 130 mutations for the various types of CMT/IN. HNF recognized the gap in supporting the neglected or ultra-rare subtypes of CMT/IN and took a bold step to create a robust natural history study to identify the unique phenotypes associated with each subtype where there were major gaps of information," said Joy Aldrich, HNF's Registry Coordinator.

"The collaboration with HNF and the incorporation of the CMTRN data into RDCA-DAP is a transformative step in our mission to tackle rare neurological disorders," said Alexandre Bétourné, Ph.D., Pharm.D., Executive Director for RDCA-DAP at C-Path. ***"This partnership broadens the scope of our portfolio and also facilitates an optimized understanding of complex conditions like inherited neuropathies. It's a leap forward in our journey to accelerate the development of innovative treatment approaches, offering a beacon of hope for individuals and families affected by these conditions."***

Read Full Press Release Click Here!

https://www.hnf-cure.org/grin-patient-registry/hnf-contributes-cmtrn-data-to-c-paths-rdca-dap/

Breaking News

for SORD Deficiency Patients

Therapeutic Research In Accelerated Discovery (TRIAD) partner Applied Therapeutics announced positive results from the 12-month interim analysis of Govorestat (AT-007) in the ongoing INSPIRE Phase 3 Trial in Sorbitol Dehydrogenase (SORD) Deficiency.

The INSPIRE trial was a Phase 3 double-blind placebo-controlled registrational study evaluating the effect of once-daily (QD) oral govorestat (AT-007) in 56 patients aged 16-55 with SORD Deficiency in the US and Europe.

The objective of this pre-specified, 12-month interim analysis was to evaluate early indicators of govorestat treatment effect in order to inform future regulatory discussions and support a potential New Drug Application (NDA) submission, due to the urgent need for treatment and absence of any other options for patients with SORD Deficiency.

The 12-month interim analysis was comprised of a clinical efficacy primary endpoint based on correlation of sorbitol with composite clinical outcome measures, and a pharmacodynamic (PD) biomarker primary endpoint based on sorbitol reduction.

Interim Analysis Results:

- Demonstrated statistically significant correlation between sorbitol level and the prespecified CMT-FOM composite clinical endpoint (10-meter walk-run test, 4 stair climb, sit to stand test,6-minute walk test and dorsiflexion) (p=0.05).
- Govorestat treatment provided sustained reduction in sorbitol level in patients with SORD Deficiency over 12 months of treatment, which was statistically significant compared to placebo (p<0.001).
- Govorestat treatment also resulted in a highly statistically significant effect (p=0.01) on the CMT Health Index (CMT-HI), an important patient-reported outcome measure of disease severity and well-being, which was a secondary endpoint in the study. Aspects of the CMT-HI that demonstrated a treatment effect included lower limb function, mobility, fatigue, pain, sensory function, and upper limb function.
- Govorestat was safe and well tolerated, with similar incidence of adverse events between active and placebo-treated groups.

HNF has been a proud partner in the patient recruitment of this trial through GRIN, educational webinars, focus groups, social media campaigns and dedicated **website content**. Our team continues to support Applied Therapeutics in increasing awareness of SORD Deficiency amongst the patient community and the importance of considering genetic testing for those patients suspected to have SORD Deficiency.

For more info on patient-initiated genetic testing, **click** **HNF's CMT Genie**.

To participate in CMT research, enroll in **HNF's Global Registry for Inherited Neuropathies, GRIN**.

To read full press release **click** **here**.

HNF DEVELOPS A SORD DEFICIENCY RAT MODEL WITH A SUCCESSFUL PHENOTYPE OF CMT.

At the prestigious University of Miami Miller, School of Medicine.

SORD-deficient rats develop a motor-predominant peripheral neuropathy unveiling novel pathophysiological insights and recent publication in BRAIN March 2024. **Click Here!**

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- HNF Centers of Excellence
- The CMT Genie
- Join Team CMT
- Patient Resources
- CMT Biobank
- GRIN

Global Registry for Inherited Neuropathies (GRIN):

A Patient-Powered Registry Boosts the Study of Charcot-Marie-Tooth (CMT) Disease

CMT is a genetic, degenerative neuromuscular disease that affects 1:2500 in the US and 2.5 million worldwide—many are still undiagnosed—and currently, there is no cure. CMT is progressive, and over time, muscles in the feet, legs, and hands lose strength. Muscles waste away and cause atrophy leading to mobility issues. It can have a serious impact on vision, hearing, breathing, speech, and swallowing in extreme cases. Some patients experience hip dysplasia, scoliosis, and/or blindness.

MISSION:
As part of TRIAD, the Global Registry for Inherited Neuropathies (GRIN) was established as a patient registry and research consortium to advance knowledge of patient records, analyze patient-reported data, collect genetic reports and clinical CMT validated scales (CMTPeds, CMTInfS). The data has been instrumental in identifying the burden, diagnostic journey and prevalence of CMT.

The Hereditary Neuropathy Foundation mission is to increase awareness and accurate diagnosis of Charcot-Marie-Tooth (CMT) and related inherited neuropathies, support people living with CMT and their families with critical information to improve quality of life, and fund research that will lead to treatments and cures.

www.hnf-cure.org