CAMBRIDGE, MASSACHUSETTS, FRIDAY, NOVEMBER 3, 2017

Patient-Centered

CHARCOT-MARIE-TOOTH/
HEREDITARY NEUROPATHY PRESSURE PALSIES
PAIN SUMMIT

hnf-cure.org
401 Park Avenue South, 10th Floor
New York, NY 10016
Thank you to PCORI

The Hereditary Neuropathy Foundation (HNF) would like to thank the Patient-Centered Outcomes Research Institute (PCORI) for nominating HNF for the PCORI’s Eugene Washington PCORI Engagement Award - 7238 titled “Patient-Centered CMT/HNPP Pain Summit.”

HNF is grateful for the support of PCORI for our 2nd Annual Summit which includes patients with Charcot-Marie-Tooth (CMT)/Hereditary Neuropathy Pressure Palsies (HNPP) and other related inherited neuropathies (IN), caregivers, clinicians, researchers, funding agencies, payors, leading pain experts and industry to come to an understanding of pain in the CMT/HNPP community, including the impact on quality of life.

The Summit you are attending today was partially funded by this award and was instrumental in the success of the Summit. We would also like to give a special thank you to Pharnext, Acceleron Pharma, Pfizer, Flex Pharma, Ortho Rehab Designs and Neurology Reviews for their continued support of the HNF’s programs and dedication to working to bring therapies to our patient community.
It is with pleasure that I welcome you to the Annual Patient-Centered CMT/IN Summit. This year we are highlighting chronic pain, and wanted to be sure to include a specific form of inherited neuropathy, hereditary neuropathy pressure palsies (HNPP), a type of CMT that often doesn’t get attention because it doesn’t have the name CMT in it.

We are thrilled to have you join us for the 2nd Annual Patient-Centered Summit.

HNF recognizes the importance of meaningful participation of patients and caregivers to engage research related to pain. The goal of today will be to come to an understanding of pain in the CMT/HNPP Community, including the impact on quality of life. Participation in the Summit will help patients to better describe and identify their pain and therapeutic approaches.

What makes this Summit so special is that the innovative content was developed in collaboration with numerous patient advocates, caregivers, clinicians, researchers, funding agencies and industry with one unified goal in mind: giving the patient a voice in discussing barriers and opportunities impacting their treatment.

You will have the chance to hear the latest in research developments, participate in panel discussions with clinicians, and voice your needs and priorities in regards to CMT/HNPP research and treatments.

Uniting everyone in the CMT/HNPP community will guide research to be more patient-centered, leading to lifestyle guidelines and treatments for those living with CMT/HNPP.

Whether you are a patient, caregiver, medical provider, or industry stakeholder, all of us are accountable for treating pain. Success depends on working together for a united purpose. This Summit is a pivotal event in the CMT/HNPP community, where you can actively participate and truly make a difference.

HNF believes that bringing all groups TOGETHER can influence research to be more patient-centered, useful, trustworthy, and ultimately lead to lifestyle guidelines and treatments for CMT/HNPP, making a difference in your life TODAY.

This Summit is designed BY patients and FOR patients. We NEED people with CMT/HNPP to participate and make this Summit a S-U-C-C-E-S-S!

Warm regards,

Allison Moore
Founder and CEO
Hereditary Neuropathy Foundation
Friday, November 3, 2017

7:30am – 8:00am Registration
8:00am – 9:00am Breakfast
9:00am – 9:05am Opening Remarks
Sean Ekins, MSc, PhD, DSc, Chief Science Officer-HNF and Master of Ceremonies
9:05am – 9:15am Welcome
Allison Moore, Founder/CEO, HNF
Result of CMT Pain Telephone Survey
Robert Moore, CEO, Footprint Digital, LLC
9:15am – 9:45am Keynote Address: “Chronic Pain and the Importance of Social Networks”
Pamela Katz Ressler, RN, MS, HNB-BC is the founder of Stress Resources in Concord, Massachusetts, a firm specializing in building resilience for individuals and organizations through tools of connection, communication and compassion.
Special Guest: John Novack, Director, Communications, INSPIRE
9:45am – 10:15am “Pain Scientific Review”
Clifford Woolf, MD, PhD, Professor in Neurology and Neurobiology-Program in Neuroscience, Harvard University
10:15am – 10:25am Q&A with Presenters
10:25am – 10:40am Coffee Break with Exhibitors
10:40am – 12:30pm General Session with Clinical Experts
“Chronic Pain Diagnosis and Treatment”
Todd Levine, MD, Phoenix Neurological
“CMT/HNPP 101”
Florian P. Thomas, MD, MA, PhD, MS, Founding Chair and Professor, Department of Neurology Hackensack University Medical Center and Seton Hall-Hackensack Meridian School of Medicine; Director, Hereditary Neuropathy Foundation Center of Excellence; Director, Multiple Sclerosis Center
“Using Physical and Occupational Therapy for Pain Management”
James Nussbaum, PT, PhD, SCS, EMT, Clinical and Research Director of ProHealth & Fitness PT OT
“Bracing Options for CMT Pain Management”
Mitchell Warner, CPO, Ortho Rehab Designs
Q&A with Presenters
Panel: “Effective Holistic and Alternative Pain Management”
Bob Twillman, PhD, Executive Director, Academy of Integrative Pain Management (AIPM)—Moderator
Pamela Katz Ressler, RN, MS, HNB-BC, Founder, Stress Resources
Kristin Gelzinis, LMSW, HNF Patient Advocate
Q&A with Panelists
12:30pm – 1:30pm  Lunch and Networking  
**Lunch Speaker** “National Pain Policy Overview”  
**Bob Twillman, PhD**, Executive Director, Academy of Integrative Pain Management (AIPM)

1:30pm – 2:00pm  General Session  
“Update on NORD’s I AM RARE Registry and Charcot-Marie-Tooth Research Network (CMTRN)”  
**Allison Seebald**, Research Programs Associate, National Organization for Rare Diseases (NORD)  
**Study Review:** “Patient Assisted Intervention for Neuropathy: Comparison of Treatment in Real Life Situations (PAIN-CONTROLS)”  
**Mamatha Pasnoor, MD**, Associate Professor-Neurology, University of Kansas Medical Center

2:15pm – 3:30pm  Workshops  
A. “The CMT/HNPP Patient Experience Workshop”—Samberg Salon IV  
**Libby Bradshaw, DO, MS**, Assistant Professor of Public Health and Community Medicine; Academic Director, Pain Research, Education and Policy (PREP) Program, Tufts School of Medicine

B. “Communicating Your Pain Workshop”—Samberg Dining Room 5  
**Gwenn Herman, LCSW-C, DCSW**, Clinical Director and Founder, Pain Connection (a U.S. Pain Foundation program)

3:30pm – 4:00pm  Coffee Break with Exhibitors

4:00pm – 5:30pm  General Session – Research and Clinical Trials Update  
“Update on HNF Initiatives”  
**Allison Moore**, Founder/CEO, HNF  
“Report from Patient Roundtables”  
**Libby Bradshaw, DO, MS**, Assistant Professor of Public Health and Community Medicine; Academic Director, Pain Research, Education and Policy (PREP) Program, Tufts School of Medicine

“Update on Current Clinical Trials for CMT and Research Update on HNPP”  
**Sean Ekins, MSc, PhD, DSc**, CEO, Collaborations Pharma  
**René Goedkoop, MD**, Chief Medical Officer, Pharnext  
**Kenneth Attie, MD**, VP-Medical Research, Acceleron Pharma  
**Thomas C. Wessel, MD, PhD**, Chief Medical Officer, Flex Pharma

Q&A with Presenters/Closing Comments

5:30pm – 7:30pm  Reception
A special thanks to this year’s Sponsors, Partners and Collaborators

Acceleron
BioFreeze
Bombas Socks
Dorothy J. Fairman Travel Scholarship
Flex Pharma
Neurology Reviews
Ortho Rehab Designs
PenAgain
Pfizer
Pharnext

Thank you to the Planning Committee members who helped shape the content, identify speakers and panelists, and develop a comprehensive program to share with you:

Joy Aldrich
Director, Advocacy, HNF
Matthew Downing
Secretary, HNF
Kristin Gelzins
Patient Advocate
Amy Goldstein
Director, State Pain Policy Advocacy Network, Academy of Integrative Pain Management
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Founding Chair & Professor, Department of Neurology, Hackensack University Medical Center and Seton Hall-Hackensack Meridian School of Medicine
Tina Tockarshewsky
Director, Medical and Public Affairs, HNF

So please join in,
make new friends, create new collaborations, build new partnerships,
but most importantly, Be Inspired!

On behalf of the entire Hereditary Neuropathy Foundation Team and our Planning Committee, we welcome you to Cambridge! We are glad you are able to attend.
Sean Ekins, PhD, DSc

Opening Remarks

9:00am – 9:05am

Sean graduated from the University of Aberdeen; receiving his M.Sc., PhD in Clinical Pharmacology and DSc in Science. He then went to the USA and was a postdoctoral fellow at Lilly Research Laboratories, worked as a senior scientist at Pfizer and then Eli Lilly. He went on to join startup companies as Associate Director of Computational Drug Discovery at Concurrent Pharmaceuticals Inc. (now Allergan) and Vice President of Computational Biology at GeneGo (now Thomson Reuters).

Sean is currently co-founder and CEO at Phoenix Nest Inc. focused on Sanfilippo Syndrome and Founder and CEO of Collaborations Pharmaceuticals, Inc. which is focused on using machine learning approaches for rare and neglected disease drug discovery. He is also the CSO of the Hereditary Neuropathy Foundation, on the SAB of the Pitt Hopkins Research Foundation and Adjunct Professor at several US Universities.

Since 2005 he has been awarded 14 STTR/ SBIR grants, 1 R21, 1UH2 and 1 R01 NIH grants as well as performs as a consultant on many others.

He has authored or co-authored and peer reviewed papers, book chapters, and edited 5 books. For over 21 years he has been at the forefront of using commercial and in-house developed software in drug discovery research. He has a passion for finding new collaborators and helping rare disease families find a pathway to a treatment for their children.

Allison Moore

Welcome

9:05am – 9:10am

Allison Moore founded the Hereditary Neuropathy Foundation (HNF) in 2001 to improve the quality of life for people living with Charcot-Marie-Tooth (CMT) and related inherited neuropathies (IN) while also seeking to treat/cure these progressive neurological disorders. In 1995, Moore herself experienced the severe and immediate onset of CMT-associated symptoms after receiving a CMT contraindicated drug to treat a serious, non-CMT medical condition.

It took two years before she was officially diagnosed with CMT1A in 1997, after a long and frustrating diagnostic process. A lifelong skier and exercise enthusiast before the onset of her CMT, today she cannot walk without leg braces and has severe muscle atrophy, bilateral foot drop, and weakness and deformity in her hands. Her personal struggle with getting a proper diagnosis, combined with the lack of public awareness about CMT, led her to start the Hereditary Neuropathy Foundation (HNF).

Moore’s mission in founding HNF was to encourage the development of treatments and cures for the 90+ genetic mutations causing progressive inherited neurological diseases. HNF has made great strides in building awareness and securing funding to support their Therapeutic Research In Accelerated Discovery (TRIAD) program, a collaborative effort with academia, government and industry, to develop treatments and cures for CMT/IN.

Moore’s unique personal experiences drive her commitment to the importance of putting patients first. HNF has developed a focus on patient engagement methods, with an emphasis on identifying the gaps in patient-reported outcomes (PROs) that are hindering patient care, standard of care guidelines, early and accurate diagnosis, therapy development and improved clinical outcome measures and endpoints to support clinical trials. HNF continues exploring
ways to implement the lessons learned from HNF’s first Summit, and plans to leverage the findings from the 2017 Summit to improve upon the under-reported impact of chronic pain on patients’ quality of life.

SPEAKER

Robert Moore

Results of CMT Pain Telephone Survey

9:10am - 9:15am

Robert Moore, husband to Allison Moore, CEO and Founder of HNF, will be discussing the results of HNF’s Telephone Survey on Hereditary Neuropathy Pain at this year’s Summit. HNF created this survey to address the gaps in pain management for hereditary neuropathy patients.

Watching Allison have trouble walking, or performing simple tasks such as buttoning a shirt or opening a bottle, has given Robert a very different perspective for anyone living with chronic pain. He has witnessed Allison go through countless visits with medical professionals that either dismissed or ineffectively managed her pain symptoms. Raising awareness of how patients experience and the challenges they face managing chronic pain will unite HCP’s, researchers, and industry to create effective pain management programs for all hereditary neuropathy patients.

The pain survey asked questions regarding how hereditary neuropathy patients experience and manage pain on a daily basis. The data collected is part of HNF’s mission to pursue quantitative and qualitative patient-centered outcomes to better define patient pain and make HCP’s, researchers, and industry aware of the challenges CMT patients face when managing chronic pain.

KEYNOTE ADDRESS

Pamela Katz Ressler, RN, MS, HNB-BC

Chronic Pain and the Importance of Social Networks

9:15am – 9:45am

Pamela Katz Ressler, RN, MS, HNB-BC is the founder of Stress Resources in Concord, Massachusetts, a firm specializing in building resilience for individuals and organizations through tools of connection, communication and compassion. Pam is a frequent, sought after speaker to local, national and international audiences on strategies of resilience. She is a faculty member at Tufts University School of Medicine in the Pain Research, Education and Policy Program, serves on the Executive Board for Medicine X at Stanford University and is an appointed member of the Consumer Health Advisory Board of the Massachusetts Health Quality Partners.

A graduate of the University of Michigan and Tufts University School of Medicine’s Pain Research Education and Policy Program, Ms. Ressler’s work in resilience is grounded in both the science and the art of health and human connection. She has received additional training at both Harvard University and the Center for Mindfulness in Medicine, Health Care and Society at the University of Massachusetts Medical School.

Pam is a featured contributor to WBUR’s Cognoscenti pages, an author with various print and online journals, and has been interviewed extensively on her research on resiliency and social media in healthcare. Her academic research on the intersection of social media and healthcare has earned her the distinction as one of the country’s thought leaders in the field. Her research on Communicating the Experience of Chronic Pain and Illness through Blogging was published in the Journal of Medical Internet Research in 2012, and remains frequently cited in academic literature. In 2015, Ms. Ressler was selected as one of 30 collaborators from the US

Ms. Ressler’s consulting work with businesses, schools, and healthcare organizations is ongoing and expanding as leaders seek strategies of sustainable resilience in our fast paced society. Cultivating Resilience, Pam’s upcoming book, will be published in 2018.

SPECIAL GUEST

John Novack

Pain Text Poll Survey

John oversees communications for Inspire, a healthcare social network of over one million patients and caregivers interacting in 200 online communities, including the HNF online support group. John served on the Association of Health Care Journalists’ board of directors. He was group publisher at HCPro, a healthcare regulatory publishing and training company. He also worked in communications and market development roles with safety software company Quantros, and EBSCO Publishing.

John has a degree in Psychology from Salem State University and a master’s degree in Journalism from Boston University. He is a former board member and current committee member of SCAD Alliance, a rare disease patient advocacy organization.

SPEAKER

Clifford J. Woolf, MD, PhD

Pain Scientific Review

9:45am – 10:15am

Dr. Clifford J. Woolf works on pain and on the regeneration and degeneration of the nervous system, with a particular focus on neurological disease modeling and drug screening in patient iPS cell derived neurons. He was born in South Africa, where he earned his MD and PhD degrees. He moved to London in 1979 and became Professor of Neurobiology at University College London. In 1997 he was recruited by the Massachusetts General Hospital, and Harvard Medical School (HMS) to serve as the first Richard J. Kitz Professor of Anesthesiology Research at HMS. In 2007, he was appointed Principal Faculty Member of the Harvard Stem Cell Institute and in 2010, was named director of the F. M. Kirby Neurobiology Center at Boston Children’s Hospital, and became Professor of Neurology and Neurobiology at HMS. Dr. Woolf is deputy director of the Intellectual Developmental Disability Disorders Center at Boston Children’s Hospital and co-director of the neuroscience program of the Harvard Stem Cell Institute.

Over his career Dr. Woolf has received many honors and prizes. He was awarded the Gill Distinguished Scientist award and the Reeve-Irvine medal this year and in 2015, the Kerr award from the American Pain Society, a Founders Award from the American Academy of Pain Medicine and became an honorary fellow of the Irish College of Anesthetists. He was awarded the Magnes medal in Israel in 2013, and selected to deliver the FE Bennett Memorial Lecture by the American Neurological Association in 2012. He was awarded a Javits Award from the NINDS at the NIH in 2011, delivered the Schmidt lecture at MIT in 2011, the Bonica Lecture for the International Society of the Study of Pain in 2010, was Visiting Professor at Columbia University in 2009, and received the Wall Medal from the Royal College of Anesthetists in the UK also in 2009. In 2015, Dr. Woolf was appointed to the Board of Scientific Councillors of the National Institute of Neurological Diseases and Stroke.
General Session 10:40am - 12:30pm

SPEAKER

Todd Levine, MD

Chronic Pain Diagnosis and Treatment

Dr. Levine is a graduate of Duke University Medical School. He completed his internship in Internal Medicine at Jewish Hospital in St. Louis. His neurology residency was at Barnes Hospital in St. Louis. He then completed a two year fellowship in neuromuscular disease at Washington University in St. Louis. He has dual board certification in Neurology as well as Electrodiagnostic Medicine.

Dr. Levine has been a member of Phoenix Neurological Associates since January of 1999. Since moving to Phoenix he has developed a subspecialty practice in neuromuscular diseases. He is the founder and director of the Phoenix Neurological ALS clinic. He is an adjunct Professor of Neurology at Kansas University. He lectures both nationally and internationally on diseases of the nerve and muscle. He also conducts extensive research and continues to publish his findings in numerous peer-reviewed journals.

SPEAKER

Florian P. Thomas, MD, MA, PhD, MS

CMT/HNPP 101

Florian P. Thomas, MD, MA, PhD, MS, serves as Professor and Founding Chair of the Department of Neurology at Seton Hall-Hackensack Meridian School of Medicine. At Hackensack University Medical Center he is the Chair of the Neuroscience Institute and Founding Chair of the Department of Neurology, as well as Director of the Multiple Sclerosis Center and the Hereditary Neuropathy Foundation Center of Excellence.

He is board certified in spinal cord medicine, neurology, and neural repair and rehabilitation, and fellowship trained in neuroimmunology and neuro-AIDS. From 1995 until 2016, he served as a tenured Professor of Neurology, Adjunct Professor of Physical Therapy, and Associate Professor of Molecular Virology, at St. Louis University, where he directed the National MS Society Multiple Sclerosis Center, the Hereditary Neuropathy Foundation Center of Excellence, and the Clinical Research Unit of the Departments of Neurology and Psychiatry. He now serves there as Professor Emeritus.

From 2004 until 2011, he served as Director of the Spinal Cord Injury Service, and from 2000 to 2015 as the Director of the Regional MS Center of Excellence at the St. Louis VA Medical Center, where he also founded the Pain Rehabilitation Service.

Dr. Thomas has expertise in MS, spinal cord injury, neuromuscular diseases, pain rehabilitation, sexual health, neuro-AIDS, immunology of the peripheral/central nervous systems, and neurogenetics. His research focuses on MS, spinal cord injury rehabilitation and the treatment and genetics of hereditary neuropathies. Since 1987 he has engaged in clinical and basic science research on Charcot-Marie-Tooth Diseases, identified a novel CMT gene in 2006 and acted as site or national principal investigator for two CMT treatment trials.
Dr. Thomas is an elected fellow of the AAN and the ANA. He chairs the Advisory Board of the State of Missouri Spinal Cord Injury/Disease Research Program. He is editor-in-chief of the Journal of Spinal Cord Medicine and is a member of several other editorial boards.

He serves on advisory boards for the Hereditary Neuropathy Foundation, the National MS Society, and the St. Louis Center for Independent Living “Paraquad”. Together with a patient advocate he founded and led, for 15 years, the St. Louis Chapter of the Charcot-Marie-Tooth Association, for which he also chaired the Grant Committee and served on the Medical Advisory Board.

**SPEAKER**

James Nussbaum, PT, PhD, SCS, EMT

Using Physical and Occupational Therapy for Pain Management

James is the Clinical and Research Director of ProHealth & Fitness PT OT, a physical and occupational therapy organization in New York. He founded the organization in 2000 in an effort to provide skilled rehabilitation services to patients with all levels of abilities and disabilities. His career was fueled by his experiences as a physical therapy patient as well as his time working for Johnson and Johnson as a health and fitness specialist. James returned to school after working at J & J and graduated from Touro College with a Masters of Science degree in Physical Therapy, later earned a PhD in Pathokinesiology, and then received board certification as a Sports Certification Specialist.

James sits on numerous clinical and academic advisory boards including the Hereditary Neuropathy Foundation, the Charcot Marie Tooth Association, Touro College’s Physical Therapy Program, served as the Co-Director of Research at TRIARQ, is the Clinical Coordinator of Clinical Education partnering with over a dozen APTA accredited PT Doctoral Programs, and is an active member of the NYS Task Force on Fall Prevention. He currently is a clinical instructor for numerous Physical Therapy schools, and teaches Human Gait, Biomechanics and Kinesiology at Yeshiva University’s Stern College.

James has been an active volunteer NYS DOH Emergency Medical Technician crew chief, and a First Aid, Water Safety, and American Red Cross CPR instructor for almost 30 years. He has provided emergency game coverage for elementary, high school and college athletic events. James’ clinical and research focus has been on the effects of innovative therapy interventions in medically complex patient populations. He has numerous ongoing IRB approved clinical trials, collaborating with many NYC physicians, hospitals, and agencies. James has lectured and presented his research around the world, and has a particular interest in finding ways to enhance and document patient improvements in activities of daily living, mobility, balance, function, and most importantly quality of life.

**SPEAKER**

Mitchell Warner, CPO

Bracing Options for CMT Pain Management

Mitchell Warner graduated from the New York University Post Graduate Medical School’s program in Orthotics and Prosthetics, and is American Board Certified in both prosthetics and orthotics.

Established in 1991, Ortho Rehab Designs is nationally recognized for the Helios® Orthotic System. The Helios® orthosis is a custom energy loading graphite composite brace. Easy to put on and take off, the unique design is an incremental improvement over existing bracing systems. It uses Triplanar control to correct and stabilize the joints of the foot and ankle.

Mitchell holds a patent for the custom energy storing Helios® bracing system designed to address CMT patient’s needs. He has developed several different types of energy storing devices for pediatrics and adults. These include the Double Helix®, Helios®, Helios® Excel, and Helios GX® orthoses.

The Helios® was used to test CMT patients in a gait study conducted at UNLV with the support of HNF and Mitchell Warner.
Functional and dynamic response characteristics of a custom composite ankle-foot orthosis for Charcot-Marie-Tooth patients was published in Gait & Posture Journal in 2013.

Mitchell wears a Helios® orthosis on his right leg to specifically treat chronic joint pain due to joint deformation and to control diminished foot and ankle functions.

Mitchell Warner’s practice specializes in treatment for CMT and he treats patients nationally and internationally.

**SPEAKER/MODERATOR**

Bob Twillman, PhD

Effective Holistic and Alternative Pain Management

Bob is the Executive Director of the Academy of Integrative Pain Management (formerly the American Academy of Pain Management). In that capacity, Dr. Twillman is responsible for guiding the organization in its efforts to promote an integrative approach to managing pain. He formerly served the Academy for four years as its Director of Policy and Advocacy. Dr. Twillman received his PhD in Clinical Psychology at the University of California in Los Angeles, and maintains a volunteer faculty appointment as Clinical Associate Professor of Psychiatry and Behavioral Sciences at the University Of Kansas School Of Medicine in Kansas City, KS. Prior to working for the Academy, Dr. Twillman was a full-time faculty member at the University of Kansas Medical Center, where he founded and directed the inpatient pain management program and was a co-founder of the hospital’s Palliative Care Team. Dr. Twillman also served for many years as Chair of the Prescription Monitoring Program Advisory Committee for the Kansas Board of Pharmacy.

**PANELIST**

Kristin Gelzinis, LMSW

I received my Masters in Social Work from Stonybrook University in 2008. I have always wanted to work with people who have disabilities or chronic conditions, becoming a social worker and connecting with HNF has allowed me to do just that. Together we launched the CMT-Connect initiative, which has been an alternative holistic way of exploring different avenues of coping, living, and thriving with CMT.

I have CMT 4C and my six year-old son has inspired me to be the best I can be. I have stayed active in martial arts and bike riding after many years of being told by my doctors that I would be best off in a wheelchair to preserve my joints and legs as much as possible.

I feel confident bringing my experience and education into my discussion at the Summit. Not only do I experience chronic pain everyday, I have worked with people who experience chronic pain on a daily basis. I recently have started a mindfulness program to add to my toolbox of coping with pain. I firmly believe that it’s a multi-pronged issue and should be addressed as such. No one way is going to be the only way to handle pain issues. I think with the right medication and skills, pain can be managed and a person’s life can be more meaningful.
General Session 1:30pm - 2:00pm

SPEAKER

Allison Seebald

Update on NORD’s I AM RARE Registry and Charcot-Marie-Tooth Research Network (CMTRN)

Allison Seebald is a Research Programs Associate at the National Organization for Rare Disorders (NORD), where she supports the I AM RARE Registry Program, as well as other initiatives such as NORD’s research grants program. Allison believes strongly in the power of natural history data and patient-centered outcomes research and is excited to be discussing the importance of natural history studies at the 2017 HNF Patient-Centered CMT/HNPP Pain Summit.

Allison’s interest in the nervous system began during her undergraduate studies at Princeton University, where she majored in molecular biology with a certificate in neuroscience, and increased further during her traineeship in developmental disabilities at Robert Wood Johnson Medical School. Prior to joining NORD, Allison spent several years working in laboratories at Cornell University researching the mechanisms of cancer and genetics.

SPEAKER

Mamatha Pasnoor, MB.BS/MD

Study Review: Patient Assisted Intervention for Neuropathy: Comparison of Treatment in Real Life Situations (PAIN-CONTRoLS)

Dr. Pasnoor received her Bachelor of Medicine and Bachelor of Surgery (MB.BS/MD) degree at Osmania Medical College, Andhra Pradesh, India. She completed an internship in medicine at Grand Rapids Medical Education and Research Center, Michigan State University, followed by a residency in Neurology and a fellowship in Clinical Neurophysiology with focus on neuromuscular diseases at the University of Kansas Medical Center. After completing her fellowship in 2006, she joined the faculty at the University of Kansas Medical Center.

Dr. Pasnoor is certified by the American Board of Psychiatry and Neurology, American Board of Neuromuscular Medicine and American Board of Clinical Neurophysiology. She also serves as co-director of the University of Kansas Neuropathy Center, along with Dr. Mazen Dimachkie, associate program director for the Neurology Residency Program and associate program director for Clinical Neurophysiology and Neuromuscular Medicine.

Dr. Pasnoor’s practice focuses on neuromuscular diseases and general neurology. Dr. Pasnoor has published in areas of peripheral neuropathies, myasthenia gravis, motor neuron disease and myopathies. She is actively involved in several clinical research programs including diabetic and other neuropathies, myasthenia gravis, myopathies and motor neuron diseases.
Libby Bradshaw, DO, MS
The CMT/HNPP Patient Experience Workshop

2:15pm – 3:30pm (Samberg Salon I/T)

Ylisabyth (Libby) S. Bradshaw, DO, MS is Academic Director for the Pain Research, Education and Policy Program in the Public Health program at Tufts University School of Medicine. Dr. Bradshaw teaches students within this and other Public Health and Professional Degree Programs, as well as pre-clinical and clinical medical students.

She has led innovations in medical and pain education for over 30 years. Her clinical practice in family medicine and then, emergency medicine led to expand her early interests in injury control and violence prevention to include public health, and behavioral and organizational change. Personal experience with injury expanded her interests to pain and disability. She is an innovative educator, encouraging active learning and engagement, and works to overcome barriers to understanding pain and suffering.

Dr. Bradshaw is a past president of the Massachusetts College of Emergency Physicians; served three terms on the Public Health Committee of the American College of Emergency Physicians; and continues to serve on the Massachusetts Medical Society’s Committees on Public Health and on Violence Intervention and Prevention and on the Massachusetts Pain Initiative Policy Council. Recently she participated in the Massachusetts Governor’s Working Group to develop shared competencies in pain for medical students statewide.

Gwenn Hope Herman, LCSW-C, DCSW
Communicating Your Pain Workshop

2:15pm – 3:30pm (Samberg Dining Room 5)

Gwenn graduated from the University of Oklahoma, receiving her Master of Social Work in 1978, and graduated from the State University of New York at Buffalo where she received her Bachelor of Science from the School of Social Work in 1975. She is a Licensed Certified Clinical Social Worker and Diplomate in Clinical Social Work, with over forty years of experience in the field of alcoholism and addictions, physical and sexual abuse, dual-diagnosis, and twenty years of experience in the field of chronic pain.

She was the Founder and Executive Director of Pain Connection® – Chronic Pain Outreach Center, Inc. from April, 1999-May, 2016. She co-authored the book “Making the Invisible Visible: Chronic Pain Manual for Health Care Providers’ with Mary French, RN, MSW, LCSW-C, which is used in trainings and “Filling the Gaps in Pain Care” sessions. In May 2016, Pain Connection® joined the U.S. Pain Foundation, Inc. and serves as their direct services component. Pain Connection® is now a program of the US Pain Foundation. She is currently licensed in Maryland and Arizona and has a small private practice in Tucson at www.gwennherman.com.

Gwenn moved to Tucson, Arizona a little over a year ago due to her chronic pain. The dry heat and sunshine has helped to lower her fibromyalgia levels. She lives in Tuscon with her husband, Malcolm, their cat Harley, and Rocky their rescue dog. They live near the Catalina Mountains and love the views and wildlife around them.
General Session – Research and Clinical Trials Update 4:00pm - 5:30pm

René Goedkoop, MD

Status of The Pivotal Phase III Study (PLEO-CMT) Assessing The Efficacy Safety of PXT3003 in The Treatment of Adult Patients With Charcot-Marie-Tooth Type 1A

In his role, he is responsible for the clinical product development of PXT3003 for CMT1A and other products in development for neurodegenerative diseases, and regulatory affairs.

Over 25 years of experience in worldwide clinical product development with emphasis on “first in class” therapeutic solutions for inflammatory, neurologic and cardiovascular disease, cancer, including experience in the field of medical devices and cell-based technologies. Clinical target indications included rare and ultra-rare genetic diseases.

He worked in start-up and large companies, including Apoix, Modex, Serono, Eli Lilly and Centocor at a global level. He has directly interacted with key regulatory agencies. In conjuncture, he has worked as an independent expert clinical development strategy consultant for a variety of biotech companies, and has conducted due diligences and helped create the business plan for de novo biotech companies for Venture Capital. He also was on the board of directors at Neovacs. He received his MD at the University of Amsterdam followed by a residency in cardiopulmonary surgery.

Kenneth Attie, MD

An Ongoing Phase 2 Study Evaluating The Safety, Efficacy, and Pharmacokinetics Of ACE-083 in Patients with CMT1 and CMTX.

Dr. Attie has devoted over 25 years to developing investigational drugs, primarily for growth disorders and neuromuscular diseases. He joined Acceleron in November 2009, as VP of Medical Research. For Acceleron, he has been medical lead for studies with TGF-β superfamily investigational drugs for anemias due to thalassemia and myelodysplastic syndromes and for neuromuscular disorders. Acceleron has brought drug candidates into phase 2 studies for indications including Duchenne and FSH muscular dystrophies and CMT. He has held similar leadership positions at Altus Pharmaceuticals and Insmed Inc., where he helped gain FDA approval of an IGF-1 product. Prior to that, Dr. Attie worked for 12 years at Genentech, Inc., where he did research related to the TGF- superfamily and was responsible for several phase 1-4 studies of recombinant human growth hormone and for obtaining marketing authorization for rhGH in chronic renal insufficiency, Turner syndrome, adult GH deficiency, and idiopathic short stature, as well as for a long-acting GH product. Dr. Attie was Assistant Clinical Professor of Pediatrics at the University of California, San Francisco Medical Center, is board-certified in Pediatrics and Pediatric Endocrinology, and has over 50 publications in peer-reviewed journals. Dr. Attie received a B.A. in Music from the University of Michigan, Ann Arbor, and an M.D. from New York University Medical Center.
SPEAKER

Thomas C. Wessel, MD, PhD

Discussion of the study to assess the safety and effectiveness of FLX-787 in CMT patients with muscle cramps in the United States

Tom Wessel is a Board-certified neurologist with over 20 years of drug development experience in neurology and psychiatry. After completing his residency training in neurology at New York Hospital and Memorial Sloan-Kettering Cancer Center, Tom stayed on the faculty as an Assistant Professor in the Department of Neurology and Neuroscience for several years, before joining the pharmaceutical industry.

He worked on a number of CNS projects at Johnson & Johnson and headed the NDA submission team for Razadyne® (galantamine) which was approved by the FDA for the treatment of mild-moderate Alzheimer’s disease in 2001.

Subsequently, Tom served as the Senior Vice President of Clinical Research at Sepracor and led several CNS clinical development teams, including the Lunesta® (eszopiclone) development program.

More recently, he worked as the Chief Medical Officer at Acorda Therapeutics where he played a critical role in the NDA submission for AMPYRA® (dalfampridine) for walking improvement in MS, approved in January of 2010 by the FDA.

Tom is now the Chief Medical Officer at Flex Pharma, a small biotech company in Boston, focusing on therapies for cramps, spasms and spasticity. The FDA has granted Fast Track designation for Flex’s lead compound FLX-787 for the treatment of severe muscle cramps in ALS. Flex Pharma has two on-going phase2 studies for muscle cramping in motor neuron disease and CMT neuropathy in the United States.
AN ONGOING PHASE 2 STUDY EVALUATING THE SAFETY, EFFICACY, AND PHARMACOKINETICS OF ACE-083 IN PATIENTS WITH CMT1 AND CMTX

Attie KM1, Glasser CE1, Shy M2, Thomas FP3, Walk D4, D’Eon S1, Miller B1, Sherman ML1
(1) Acceleron Pharma, Cambridge, MA, (2) University of Iowa, Iowa City, IA (3) Hackensack University Medical Center, Hackensack, NJ, (4) University of Minnesota, Minneapolis, MN, USA,

BACKGROUND

ACE-083 is an investigational protein therapeutic that acts as a localized ligand trap for myostatin and other negative regulators of muscle growth. Local injection of ACE-083 into the gastrocnemius muscle of wild-type, mdx, and SOD1 mice produced dose-dependent increases in muscle mass and force without systemic effects.

In a Phase 1 single-center, double-blind, placebo-controlled dose escalation study in post-menopausal women, unilateral injections of ACE-083 into the rectus femoris (RF) or tibialis anterior (TA) muscle were generally safe and well tolerated. Mean percent changes from baseline in total muscle volume of the injected muscle were +14.5% in the RF and +8.9% in the TA at the highest dose administered with minimal changes observed in the contralateral side and placebo-treated subjects. Frequent related AEs (≥15%) included injection site pain, pain in extremity, injection site discomfort, and muscle twitching, with similar incidence in ACE-083 and placebo-treated groups. All AEs were grade 1-2 and reversible.

Together, these preclinical and clinical results support further studies of ACE-083 in myogenic and neurogenic diseases with focal loss of muscle strength and function, including CMT.

METHODS

Study A083-03 (NCT03124459) is an ongoing, multicenter, Phase 2 study to evaluate the safety, tolerability, pharmacokinetics, pharmacodynamics, and efficacy of ACE-083 in patients with CMT1 and CMTX. Part 1 is open-label and will enroll up to 3 dose-escalating cohorts (6 patients per cohort); Part 2 is randomized, double-blind, placebo-controlled, and will enroll an additional 24 patients. ACE-083 will be administered bilaterally into the TA muscle once every 3 weeks for 5 doses.

A Safety Review Team will meet periodically throughout the study to review safety data and make dosing recommendations, including the recommended dose level for Part 2. Eligible patients must have genetically confirmed CMT1 or CMTX with mild-moderate weakness in ankle dorsiflexion.

Safety and tolerability will serve as the primary outcome measures for Part 1, with muscle volume and intramuscular fat fraction evaluated by MRI for Part 2. Additional outcome measures of interest include strength, motor function tests, and quality of life using CMT-Health Index questionnaire. A Phase 2 study of ACE-083 is also ongoing in facioscapulohumeral muscular dystrophy (FSHD).
PRELIMINARY RESULTS FOR CHARCOT-MARIE-TOOTH PATIENT-REPORTED SURVEY


(1) Hereditary Neuropathy Foundation, New York, USA; (2) Acceleron Pharma, Cambridge, USA; (3) University of Kansas Medical Center, Kansas City, USA; (4) University of Michigan, Ann Arbor, USA; (5) University of Minnesota, Minneapolis, USA; (6) ProHealth & Fitness New York, USA

BACKGROUND
Charcot-Marie-Tooth (CMT) disease affects roughly 1 in 2,500 individuals and is described as an inherited peripheral neuropathy primarily affecting distal muscles. Limited studies detail patient-reported impact of muscle weakness on functional activities.

METHODS
This anonymous survey was developed with input from clinical experts and patient interviews and aimed to better understand the prevalence and impact of various CMT clinical manifestations on patients’ lives. The survey was administered online to the Hereditary Neuropathy Foundation’s (HNF) patient contact database. Here we present preliminary data on patient characteristics and disease impact for 626 CMT patients collected February 17–21, 2017.

RESULTS
Respondents were mostly female (61%) and mostly from the US (74%). Median age (range) at symptom onset was 14 years (0-84 years), at diagnosis was 36 years (2-83 years), and at present was 55 years (6-89 years). The sample was representative of all CMT types (CMT1,2,3,4, and X). The most common physical and clinical manifestations of CMT were problems with balance (87%), ankle weakness/foot drop (81%), loss of feeling or abnormal sensation in the lower leg/foot (81%), and hand muscle weakness (78%). Maintaining balance, walking long distances, and climbing up and down stairs were key challenges associated with ankle weakness/foot drop. Foot drop was considered by 72% to be the primary factor contributing to falls, which averaged 2.5 falls to ground per month. Of those with foot drop, 86% had bilateral weakness. A majority of respondents (76%) used some form of assistive device for mobility, including ankle-foot orthotics/below-the-knee leg braces (40%), canes/walking sticks (34%), and custom foot orthotics/inserts (26%). The most common drug therapy included pain and anti-inflammatory medications (41%). Foot surgery was the most common surgical procedure received (24%) and toe surgery was the most common surgery considered (23%). Key symptoms that affected quality-of-life “very much” included problems with balance (65%), ankle weakness (foot drop) (62%), and fatigue (49%).

CONCLUSIONS
These data suggest a high prevalence of lower leg muscle weakness; therefore, therapies aimed at improving ankle weakness and the resulting foot drop and imbalance may be beneficial to patients’ daily functioning and quality of life.
Efficacy of PXT3003 in the treatment of adult patients CMT1A (n=80) was shown in a multicenter, randomized, double-blind, placebo-controlled phase II study (Attarian et al. 2014). PXT3003 taken 2x/day, orally, for 12 consecutive months was well tolerated and safe. Significant improvement of disability was observed for the highest tested dose, thought indicative for an early, meaningful change in disease course (meta-analysis by Mandel et al., 2015). This formed the rationale to initiate a multicenter, randomized, double-blind, placebo-controlled pivotal phase III study (ClinicalTrials.gov: NCT02579759) of PXT3003 in mildly to moderately affected CMT1A patients in December 2015. The primary objective is to assess the efficacy of 2 doses of PXT3003 compared to placebo on disability as measured by the mean change from baseline Overall Neurology Limitations Scale (ONLS) score at month 12 and 15. Furthermore, efficacy on the proportion of responders (i.e. improvement of ONLS), impairment (CMTNS-v2), functional tests (10-MWT, QMT, 9-HPT), electrophysiological parameters (CMAP, SNAP and NCV) and quality of life (EQ-5D, VAS) are secondary endpoints. Pursuant this study, patients will be eligible for a 9-month extension study, in which PXT3003 assigned patients will continue with the previously assigned dose, whereas placebo patients will be randomized to one of the two PXT3003 doses. The study is conducted in 30 investigational sites in 8 countries (EU, Canada and US). In December 2016 patient randomization was completed (n=323). The screen failure rate was 26%, as expected (437 patients were screened). The independent DSMB recommended to continue the study as planned following a safety analysis on all patients in September 2017. Preliminary baseline characteristics are based on 313 patients (data not cleaned). The study population had a mean age of 40.8±13.3 years (range 16-65; male 41.2%) of which 97.8% had a confirmed genetic diagnosis of CMT1A. The mean CMTNS-v2 was 13.9±3.09 and the mean motor nerve conduction of the ulnar nerve was 23.4±11.3 m/s. Ten patients withdrew from the study, 3 due to adverse events unrelated to study treatment. The last patient completing the study is expected in June 2018.
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