FDA Patient Listening Session

CMT-SORD

SORD Deficiency, Charcot-Marie-Tooth

August 4, 2026

HEREDITARY NEUROPATHY FOUNDATION

FDA Rare Disease Patient-led Listening Session
Meeting Summary
CMT-SORD

August 4, 2026

Organizer: Hereditary Neuropathy Foundation (HNF)

Led by: FDA Public Engagement Staff using Zoom

Total Number of Participants: 52, including representatives from:

- FDA Center for Biologics Evaluation and Research (CBER)
- FDA Center for Drug Evaluation and Research (CDER)
- FDA Center for Devices and Radiological Health (CDRH)
- FDA Office of the Commissioner (OC)
- National Institutes of Health (NIH) National Center for Advancing Translational Sciences (NCATS)
- Reagan-Udall Foundation
- The Hereditary Neuropathy Foundation

Attendees on behalf of HNF:
Allison Moore, CEO and Founder, Hereditary Neuropathy Foundation
Courtney Hollett, Executive Director, Hereditary Neuropathy Foundation
Estela Lugo, Program Development Manager, Hereditary Neuropathy Foundation
Ellen Morgan, Clinical Research Officer, Hereditary Neuropathy Foundation
Stephan Zuchner, MD, PhD, University of Miami
David Herrmann, MD, University of Rochester
Susan Bruhn, CEO, Charcot-Marie-Tooth Association (CMTA)
Filippo Genovese, European Charcot-Marie-Tooth Federation (ECMTF)
Laura McNeil, Charcot-Marie-Tooth Research Foundation (CMTRF)
Patrick Livney, Cure CMT
Brian Lin, Muscular Dystrophy Association (MDA)

Patients represented:
4 Patients
1 Parent of patient
1 Spouse of patient

FDA Attendees

Office of the Commissioner (OC) – 3 offices

- Office of External Affairs/Public Engagement Staff (*organizer*)
- Office of the Chief Medical Officer/Office of Orphan Products Development
- Office of the Chief Medical Officer/Office of Pediatric Therapeutics

Center for Biologics Evaluation and Research (CBER) – 1 office

- Office of the Center Director

Center for Drug Evaluation and Research (CDER) – 6 offices

- Office of the Center Director
- Office of New Drugs
- Office of New Drugs/Office of Neuroscience
- Office of New Drugs/Office of Neuroscience /Division of Neurology II
- Office of New Drugs/Office of Rare Diseases, Pediatrics, Urology and Reproductive Medicine/Division of Rare Diseases and Medical Genetics
- Office of Translational Sciences/Office of Biostatistics/Division of Biometrics I

Center for Devices and Radiological Health (CDRH) – 9 offices

- Office of Product Evaluation and Quality/Office of Health Technology I/Division of Health Technology I C
- Office of Product Evaluation and Quality/Office of Health Technology III
- Office of Product Evaluation and Quality/Office of Health Technology III/Division of Health Technology III B
- Office of Product Evaluation and Quality/Office of Health Technology III/Division of Health Technology III C
- Office of Product Evaluation and Quality/Office of Health Technology IV
- Office of Product Evaluation and Quality/Office of Health Technology IV/Division of Health Technology IV A
- Office of Product Evaluation and Quality/Office of Health Technology V/Division of Health Technology V A
- Office of Product Evaluation and Quality/Office of Health Technology V/Division of Health Technology V B
- Office of Strategic Partnerships and Technology Innovation/Office of Equity and Innovative Development/Division of Patient Centered Development

Non-FDA Attendees

Reagan Udall Foundation

National Institutes of Health (NIH)

- NIH/NCATS – National Center for Advancing Translational Sciences

Purpose of Meeting:
The purpose of this listening session was to provide individuals living with CMT-SORD and family members an opportunity to share their experiences directly with the FDA. Discussions focused on the symptoms that most affect daily life, treatment experiences, unmet medical needs, and perspectives on future therapies.

Summary of topics discussed:

Allison Moore provided an overview of symptoms, quality of life factors, pain, and life satisfaction based on reporting by participants with CMT-SORD in the Global Registry of Inherited Neuropathies.

Dr. Zuchner discussed the Genotype and Phenotype spectrum of Charcot-Marie-Tooth disease due to mutations in the SORD gene. Molecular testing of CMT-SORD can be complicated due to a pseudogene, SORD2P, and many laboratories do not include it in their testing. Approximately 1/300 are carriers of a pathogenic allele.

The loss of the sorbitol dehydrogenase enzyme allows sorbitol levels to increase, causing neuropathy and muscle wasting. Elevated sorbitol in blood serves as a clear biomarker that can also confirm mechanism of action when treatment is applied. A possible way to treat CMT-SORD is to use an aldose reductase inhibitor to block elevated levels of sorbitol.

Dr. Herrmann discussed the clinical phenotype, impairments, and disability in CMT-SORD patients. It appears to affect males more frequently than females, approximately in a ratio of 70:30. Symptom onset is typically in the second decade (median 15 yrs, range 12-28 yrs). Key symptoms include gait and balance difficulties, difficulty walking on heels and toes, foot deformities, muscle atrophy in the legs, and muscle atrophy in the hands in 50% of patients.

These symptoms are progressive and have a significant impact on daily living. They result in difficulties running and walking in nearly all patients, while some experience other limb-related issues. Muscle atrophy in the legs is particularly prominent in CMT-SORD, often exceeding that occurring with other common forms of CMT.

Patient #1

He is an orphan, so he doesn't know if his biological parents were affected by CMT. "Since I was orphaned, I know nothing about my parents," he said. "Did they also have CMT? Is that why they abandoned me?" He was able to run as a child, but lost the ability to play sports around 2006. His progressing symptoms then led to a mental health crisis; as he described it, "I quietly began grieving the losses, as if my early childhood had come back to take the rest from me also." By 2013, he suffered from severe muscle atrophy, but was not diagnosed until 2021.

Patient #1 participated in a clinical trial which was demanding, but also rewarding. "My coordination and balance improved, and I could fold laundry without exhaustion," he said.

“When I wanted to give up, I reminded myself that an approved treatment is the best hope I can give someone who’s currently in my teenage shoes.” Now, physical therapy helps to manage his symptoms somewhat, but does not stop disease progression. His greatest fear is debilitating fatigue that prevents him from caring for his son. He hopes there will be an approved treatment in the future that will allow him to be an active and involved dad who can help his son reach his potential, to rediscover his professional purpose, and see more of the world. “When he grows up,” he said, “I want him to remember a dad who didn’t have to choose between playing in the park or taking a shower.”

Patient #2

Her symptoms started at age 11 when she went from the fastest runner to the slowest in just 2 months. “People thought I was just being lazy, but I was fighting a war inside my body,” she said. She was diagnosed at age 20, forcing her to give up her dream of playing college softball. She has had 5 surgeries to date and physical therapy for leg tightness, but nothing has helped. She lives with a lot of pain — “I live by a quote from Muhammad Ali: Don’t count the days, make the days count. But with SORD, making the days count is a grueling uphill battle,” she said. She looks normal to others but has difficulty performing simple tasks, such as household chores or even getting up to let the dog out. “It’s the anxiety of people thinking I’m faking it because I look normal,” she said.

She knows if she does one physical activity, she will be too tired to do others that day. She uses numerous temporary fixes, such as a box of lidocaine patches per day, salt baths, IV therapy and heating pads, but really wants a way to block the sorbitol that is destroying her muscles. She hopes to find a treatment that can change her future, allowing her to work as a nurse to help others, coach a team, and live the American dream. She would tolerate anything that would help her to be pain-free. “A real treatment doesn’t just change a lab value; it changes my entire future,” she said. “It means a life defined by my hopes, not by my pain.”

Patient #3

Patient #3 was an athlete until age 13 when she was cut from the volleyball team. Her condition was suspected to be psychosomatic — “I began to wonder whether I was exaggerating, not trying hard enough, or somehow responsible for what my body could not do,” she said — so it was a relief when, after 5 years, in 2022, she received a diagnosis. She suffers from a lot of fatigue that affects her education and cognition. “There is a profound disconnect between what I am capable of and what my body will allow me to do,” she said.

During the recent heat wave in London, she was unable to sleep, which increased her falls and made it more difficult to walk; “I literally thought I was going insane,” she recalled. She longs for a treatment that can preserve her function and independence and will allow her to study for a full day and make plans to build her future. “I just want a fighting chance to remain academically engaged, socially connected, and independent,” she said.

Patient #4

Patient #4 served nearly ten years as a U.S. Army Cavalry Scout before progressive symptoms from CMT-SORD altered the course of his military career and daily life. Weakness and balance issues began during childhood, but he learned to compensate, despite numerous broken bones. He somehow was able to pass the military physical at age 17 and served while symptomatic, including deployment despite worsening symptoms, such as hand tremors, cramps and balance issues. He continued to work harder and harder, causing more injuries, leading to surgeries and ultimate discharge from the Army. All at once, he lost his calling, purpose, passion, and teammates. “I wasn’t just leaving a job,” he said. “I was losing a calling. I lost my purpose. I lost my passion. I lost the teammates who had become my family.” He wanted to work and provide for his family, but saw his body failing, which led to a mental health decline. He was diagnosed with CMT at age 37, and with CMT-SORD at age 41. “For the first time, I understood that I wasn’t failing,” he said. “My body was fighting a progressive genetic disease.”

Now, his symptoms, especially the fatigue, are overwhelming, affecting his entire family. He continues exercise, physical therapy, and cycling to help maintain some muscle and stamina. He hopes to someday have a treatment that will slow progression and lessen the fatigue, giving him some quality time. “I am not asking to become the man I was at twenty years old,” he said. “I want the opportunity to remain the husband, father, and person I am today for as long as possible … Because for someone living with CMT-SORD, preserving function is not just a medical outcome. It is preserving a life.”

Parent of Patient #5

Patient #5, now age 22, loved sports as a child, especially baseball, soccer, and basketball. When he was 13, his foot deformity, clumsiness, tripping, and unsteady gait prevented him from continuing to play ball. “Watching him struggle to run the bases or grip a bat was heartbreaking,” his mother said. “Watching your child slowly lose the piece of themselves that brought them the most joy was devastating.” Because diagnosing CMT-SORD genetically is challenging, in 2019 he was diagnosed with CIDP, a chronic autoimmune disorder. Long infusions for CIDP made him sicker, becoming overwhelming. His disease progressed faster than most. Finally, he received the correct diagnosis of CMT-SORD in 2021.

He continues to live his life, but the disease affects his whole family. As his mother put it, “He has SORD, but he is not SORD.” His mother worries about his future ability to work and be independent. “If I end up in a wheelchair, it is what it is,” he has told her — words she says are incredibly difficult to hear, “not because he lacks strength, but because he has had to come to terms with possibilities that no young adult should have to face.” A treatment for him would slow disease progression and help him to keep his independence. “Meaningful treatment does not have to mean a cure,” his mother said. “It means slowing progression. It means preserving independence … Every person deserves the chance to keep being who they are — not who their disease says they will become.”

Spouse of Patient #6

It took 46 years for Patient #6 to be diagnosed with CMT-SORD. While he desperately wanted to work and be fully present in his children’s lives, he didn’t have the physical and emotional resources to do so since he was plagued by pain and a debilitating lack of energy. His legs

became smaller and smaller over time. His wife took on the full responsibility of raising their children, which at times was frustrating for both of them. “I had long since given up asking Greg questions like these,” she said. “I had long since given up expecting him to be involved in our lives in that way.”

She pretended that the children weren’t affected by the lack of a father role model and their father’s lack of involvement in their lives, but they were. “Progression means your life gets smaller and smaller,” she said. “Not just your muscles, your life. That includes relationships.” Her son had a recurrent nightmare that the family was being chased by monsters, but their Dad could get away because he couldn’t run — in her telling, “Greg can’t run, so he couldn’t keep up, and we had to leave him behind.” CMT-SORD takes a toll on the entire family. “The next family deserves better,” she said.

Patient #6 participated in a clinical trial. While receiving the experimental treatment, symptoms of balance, strength, pain, energy, and hand tremors improved, allowing him to be a more active participant in family life. “Finally, he started to become the person he had always wanted to be,” his wife said. “In his heart, Greg is a helper and servant, but he can’t be that person without treatment.”

FDA CDRH Reviewer asked: “What level of risk are patients willing to take on to access experimental or early-stage therapies?”

Responses:

- “I would do anything to be able to get out of bed. Side effects are not important.”
- “I wouldn’t want to take much risk, but many treatments are not risky. There is just a time commitment to participate in a clinical trial”.
- “My husband would do anything to make the most of the rest of his life”.
- “I spent much of my life on the couch, and would take any risk just to have 100 good days. Give me a few great years in place of 30 years in bed”.
- “Disease progression has taken away a lot of my life. Experimental treatment risks are small compared with the potential gain. I want to be able to care for my son”.

FDA Neurology Division Director asked: “What is your experience with clinical trials?”

Responses:

- “I was in the AT trial from 2021-25 and had much more energy while I was taking the drug, instead of sleeping all of the time. I would do anything to get that drug”.
- “My son’s experience in the AT trial was incredible. He was doing so much better – less tripping, better PEG tests, etc., even though he had to take a long flight each month for the study visits. He would love to get the medication again.”

- “My husband did see improvement in the AT Expanded Access.”
- “I was initially on placebo in the AT trial, but when I went into the Extension I felt great improvement in walking, and had less fatigue. I have had more pain and fatigue since stopping the drug.”

FDA Public Engagement Staff concluded the meeting by thanking all of the speakers and sponsors, and also announced that patients and advocates can still interact with the FDA via ForPatients@fda.hhs.gov.

Disclaimer

Discussions in FDA Patient Listening Sessions are informal. All opinions, recommendations, and proposals are unofficial and nonbinding on FDA and all other participants. This report reflects HNF’s account of the perspectives of patients and caregivers who participated in the Patient Listening Session with the FDA. To the extent possible, the terms used in this summary to describe specific manifestations of CMT-SORD health effects and impacts, and treatment experiences, reflect those of the participants. This report is not meant to be representative of the views and experiences of the entire CMT-SORD population or any specific group of individuals or entities. There may be experiences that are not mentioned in this report.