



UNIVERSITY of
ROCHESTER

Neuromuscular Disease Center Quarterly Update

NMD 2024 in Review

As we reflect on the numerous achievements in 2024, it's clear that this year has been nothing short of amazing for our NMD team. The community spirit has truly shone through with inspiring walks dedicated to supporting our patients, showcasing the unwavering commitment of everyone involved. Furthermore, the groundbreaking research being undertaken is not just a testament to our dedication but also a beacon of hope for those we serve. Together, we are not just making strides; we are paving the way for a brighter future for our patients, filled with promise and potential. At the heart of our healthcare mission lies an unwavering commitment to excellence, as our dedicated providers and staff work tirelessly to deliver the highest standard of care to our patients. We extend our heartfelt gratitude to each and every one of you for your unwavering dedication and tireless efforts. Your hard work has not gone unnoticed, and it plays a crucial role in our collective success. Together, we have overcome challenges and achieved remarkable milestones, and it is your commitment and teamwork that inspire us all to reach even greater heights. Thank you for being an integral part of this journey; your contributions truly make a difference!



Staff Spotlight



This quarter, it is with great enthusiasm that we shine a spotlight on our exceptional PSR/Referral Coordinator team, whose unwavering dedication and tireless efforts play a pivotal role in ensuring our patients receive timely scheduling. This team enhances the patient experience and fosters a sense of community and trust. We are genuinely grateful for their commitment and hard work, which exemplifies the spirit of teamwork and excellence that drives our organization forward. Let us celebrate their contributions and continue to support one another in our mission to provide outstanding care!

Liz Pensgen is the supervisor to Katie Bartomioli (not pictured), Tiffanie Claeys (not pictured), Ashley Claudio (not pictured), Sarina Clinkscales, Phonemani Kongmany, Chandra Martinez, and Patty Worthy.

NMD Clinical News

Director of NMD Clinical Operations

Dr. Phil Mongiovi has assumed a new role as Director of Clinical Operations for the Neuromuscular Division. Dr. Mongiovi will continue as the Neuromuscular Medicine Fellowship Program Director. He will be instrumental in working with Liz Pensgen, Lorie Wolfanger, and clinical program leads to manage the day-to-day

operations and access for our multidisciplinary neuromuscular clinics. We would like to thank Dr. Mongiovi for accepting this role and look forward to working with him in this capacity.



Dr. Patrick Curry joins NMD

Dr. Curry attended the University of Maryland School of Medicine and moved to Rochester for his Neurology Residency training. He stayed on as a Neuromuscular Fellow before joining the Neuromuscular Division faculty this past July. Dr. Curry also serves as the Associate Director of the URM Electrophysiology Laboratory.



2024 saw the addition of Katherine Ochoa, RNP to our NMD team and also the retirements of Dr. Rabi Tawil and Ann Ford Fricke, who were with the Department of Neurology for over 30 years respectively.

Dr. Tawil's immense contributions to the NMD field were celebrated in June of 2024. Click below for more details about his amazing career.

[Dr. Rabi Tawil's Career in NMD Research and Treatment](#)

And in Research

Celebrating Decades of Advances in Myotonic Dystrophy Research

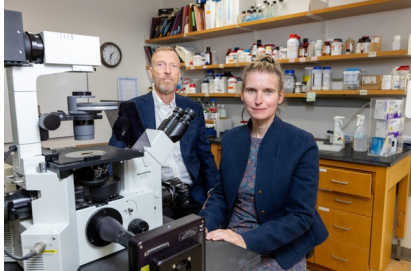
The NMD team has been at the forefront of progress in myotonic dystrophy over the past three to four decades. These



advancements have brought researchers closer than ever to developing treatments for this debilitating disease.

Click below for the full article highlighting the work of Dr. Charles Thornton and Dr. Johanna Hamel and their team.

UR Newsroom



Visiting Researcher in NMD from Rome, Italy

Laura Tufano is a neurologist and neuromuscular specialist. During her residency, her primary research interests were focused on muscle diseases, particularly Myotonic Dystrophy type 1. Her research explored muscle histology-MRI matching in muscle diseases and the development of outcome measures for Myotonic Dystrophy type 1. After completing her residency at Sapienza University of Rome in 2023, she transitioned into a PhD program to continue her research studies, under the mentorship of Dr. Matteo Garibaldi. Currently she is working on clinical research projects on Myotonic Dystrophy at the University of Rochester, where she is mentored by Dr. Johanna Hamel. She has been evaluating the test-retest reliability of remote functional assessments and sleep data collected through wearable devices as part of the REACH-DM project. In addition, Laura is focusing on Central Nervous System involvement of Myotonic Dystrophy as captured by the National Registry.

Pediatric Neuromuscular Program News



The Pediatric neuromuscular team led by Dr. Emma Ciafaloni was a top enroller for the Phase 3 EMBARK therapy trial in Duchenne muscular dystrophy (DMD) that concluded in 2024.

Results were published in Nature Medicine.

Click here to read the article: PMID: 39385046

AAV gene therapy for Duchenne muscular dystrophy: the EMBARK phase 3 randomized trial

During the study, our site was audited by FDA and did very well thanks to the incredible work from all members of the team, but most of all Patty Smith and Kim Hart our super research coordinators.

Kudos!



NMD Pediatric Team



PPMD Meeting 2025 in Sandiego, CA was a great success!



Click below to hear the interview on WXXI with Dr. Sam Mackenzie and Dr. Bo Lee regarding newborn screening for SMA and DMD.

**Understanding Gene Therapy
for Babies and Young Children**

**A Sort of
Homecoming**

Click above for a heartwarming patient story and interview with Dr. Ciafaloni.

URMC Neuromuscular Team Leading the Way Towards Therapy for Charcot Marie Tooth Disease (CMT)

Our CMT research team has coordinated a multicenter, international, NIH funded

clinical trial readiness study for CMT1A, the most common form of CMT. Participants in this study, Accelerate Clinical Trial Readiness in CMT (ACT-CMT), have completed 3 years of follow-up. The CMT research team has recently received a 1.2 million grant from the CMT Association to extend participant follow-up to 5 years and for biomarker discovery. The results from ACT-CMT are now guiding design of clinical trials for CMT. URM is currently a site for two clinical trials in CMT (NMD Synapse - CMT1 and CMT2) and Inspire (CMT-SORD), with additional clinical trials expected to open in the coming months.

Read more by clicking the below links:

PMID: 38237108 Multicenter Validation of the Charcot-Marie-Tooth Functional Outcome Measure

PMID: 39390667 Patient-reported disease burden in the Accelerate Clinical Trials in Charcot-Marie-Tooth Disease Study

NMD CMT Team and Research Coordinators



Neuromuscular Disease Center

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