



Second European CMT Specialists Conference



PUBLICATIONS/READINGS:

[Outcome measurement for assessing iNMD therapies and the effects of new drugs in clinical trials \(Davide Pareyson\)](#)
[M12 / WP 5](#)

Readings for preparing Sorted by disease, then by year (descending). Columns: Year, Disease, Outcome type, Full citation, Link:

Plenary Session 4: Clinical trials, data sharing and outcome measurement

Year	Disease	Outcome measure / scale (Type)	Full citation	Link
Charcot-Marie-Tooth disease				
2025	Charcot-Marie-Tooth (CMT)	Platform for COA and certification	Cornett KMD, Estilow T, Bray P, Mandarakas MR, Donlevy GA, Baldwin JN, Eichinger K, Finkel RS, Burns J, McKay MJ. Designing and Implementing a Web-Based Platform for Accurate and Reliable Clinical Outcome Measures and Global Certification for Evaluating Charcot-Marie-Tooth disease. J Peripher Nerv Syst. 2025 Sep;30(3):e70062. doi: 10.1111/jns.70062. PMID: 40968111; PMCID: PMC12446152.	https://pubmed.ncbi.nlm.nih.gov/40968111/
2025	Charcot-Marie-Tooth (CMT) and Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP)	Wet biomarker	Bellanti R, Keh RYS, Keddie S, Chou MKL, Misheva M, Smyth D, Baskozos G, Moodley K, Hart MS, Davies A, Reilly MM, Rinaldi S, Lunn MP. Plasma periaxin is a biomarker of peripheral nerve demyelination. Brain. 2025 Jun 23:awaf234. doi: 10.1093/brain/awaf234. Epub ahead of print. PMID: 40581376.	https://pubmed.ncbi.nlm.nih.gov/40581376/
2025	Charcot-Marie-Tooth (CMT)	Quantitative Muscle MRI	Evans MRB, Salhab HA, Sinclair CDJ, Shah S, Hanna MG, Yousry TA, Thornton JS, Morrow JM, Reilly MM. Twelve-month change in quantitative MRI calf muscle fat fraction in CMT1A predicts clinical change over 4 years. Ann Clin Transl Neurol. 2025 Apr;12(4):756-767. doi: 10.1002/acn3.52314. Epub 2025 Feb 17. PMID: 39957630; PMCID: PMC12040511.	https://pubmed.ncbi.nlm.nih.gov/39957630/
2025	Charcot-Marie-Tooth (CMT)	Nerve MRI	Zhu Y, Sun X, Fan D, Liu X. Nerve Diameter and DTI Parameters Maybe Potential Markers for Clinical Trial in Patients With Charcot-Marie-Tooth Disease Type 1A. Eur J Neurol. 2025 Jun;32(6):e70220. doi:	https://pubmed.ncbi.nlm.nih.gov/40511618/

			10.1111/ene.70220. PMID: 40511618; PMCID: PMC12163648.	
2025	Charcot-Marie-Tooth (CMT)	Wet biomarkers	Pretkálnina D, Kenina E, Gailite L, Rots D, Blennow K, Zetterberg H, Kenina V. Evaluating plasma biomarkers NfL, GFAP, GDF15, and FGF21 as indicators of disease severity in Charcot-Marie Tooth patients. Front Neurol. 2025 Jan 15;15:1490024. doi: 10.3389/fneur.2024.1490024. PMID: 39882365; PMCID: PMC11774688.	https://pubmed.ncbi.nlm.nih.gov/39882365/
2024	Charcot-Marie-Tooth (CMT)	CMT-FOM (Functional Outcome Measure) (PerfO)	Mandarakas MR, Eichinger KJ, Bray P, Cornett KMD, Shy ME, Reilly MM, Ramdharry GM, Scherer SS, Pareyson D, Estilow T, McKay MJ; for ACT-CMT Study Group; Herrmann DN, Burns J. Multicenter Validation of the Charcot-Marie-Tooth Functional Outcome Measure. Neurology. 2024 Feb 13;102(3):e207963. doi: 10.1212/WNL.00000000000207963. Epub 2024 Jan 18. PMID: 38237108; PMCID: PMC11097760.	https://pubmed.ncbi.nlm.nih.gov/38237108/
2024	Charcot-Marie-Tooth (CMT)	CMT-HI (MCID / trial-readiness) (PRO)	Rehbein T, Purks J, Dilek N, Behrens-Spraggins S, Sowden JE, Eichinger KJ; ACT-CMT Study Group; Burns J, Pareyson D, Scherer SS, Reilly MM, Shy ME, McDermott MP, Heatwole CR, Herrmann DN. Patient-reported disease burden in the Accelerate Clinical Trials in Charcot-Marie-Tooth Disease Study. J Peripher Nerv Syst. 2024 Dec;29(4):487-493. doi: 10.1111/jns.12662. Epub 2024 Oct 10. PMID: 39390667; PMCID: PMC11631656.	https://pubmed.ncbi.nlm.nih.gov/39390667/
RELEVANT OUTCOME MEASURES PUBLISHED IN PREVIOUS YEARS				
2018	Charcot-Marie-Tooth (CMT)	CMTInfS (CMT Infant Scale) (PerfO)	Mandarakas MR, Menezes MP, Rose KJ, Shy R, Eichinger K, Foscan M, Estilow T, Kennedy R, Herbert K, Bray P, Refshauge K, Ryan MM, Yiu EM, Farrar M, Sampaio H, Moroni I, Pagliano E, Pareyson D, Yum SW,	https://pubmed.ncbi.nlm.nih.gov/30476010/

			Herrmann DN, Acsadi G, Shy ME, Burns J, Sanmaneechai O. Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. Brain. 2018 Dec 1;141(12):3319-3330. doi: 10.1093/brain/awy280.	
2018	Charcot-Marie-Tooth (CMT)	CMT-HI (Charcot-Marie-Tooth Health Index) (PRO)	Johnson NE, Heatwole CR, Ferguson M, et al. The Charcot-Marie-Tooth Health Index (CMT-HI): evaluation of a disease-specific patient-reported outcome measure. Ann Neurol. 2018;84(2):225-233. doi:10.1002/ana.25282	https://pubmed.ncbi.nlm.nih.gov/30014533/
2014	Charcot-Marie-Tooth (CMT)	CMTNSv2 (CMT Neuropathy Score v2) (ClinRO)	Sadjadi R, Reilly MM, Shy ME, Pareyson D, Laura M, Murphy S, Feely SM, Grider T, Bacon C, Piscoquito G, Calabrese D, Burns TM. Psychometrics evaluation of Charcot-Marie-Tooth Neuropathy Score (CMTNSv2) second version, using Rasch analysis. J Peripher Nerv Syst. 2014 Sep;19(3):192-6. doi: 10.1111/jns.12084. PMID: 25400013; PMCID: PMC4303498.	https://pubmed.ncbi.nlm.nih.gov/25400013/
2012	Charcot-Marie-Tooth (CMT)	CMTPedS (CMT Pediatric Scale) (PerfO)	Burns J, Ouvrier R, Estilow T, Shy R, Laurá M, Pallant JF, Lek M, Muntoni F, Reilly MM, Pareyson D, Acsadi G, Shy ME, Finkel RS. Validation of the Charcot-Marie-Tooth disease pediatric scale as an outcome measure of disability. Ann Neurol. 2012 May;71(5):642-52. doi: 10.1002/ana.23572. PMID: 22522479; PMCID: PMC3335189.	https://pubmed.ncbi.nlm.nih.gov/22522479/
REVIEWS / EDUCATION MATERIAL ON CMT COA				
2025	Charcot-Marie-Tooth (CMT)	Pain management	Steve S Scherer - Webinar	https://www.youtube.com/watch?v=KELsjGPvMYQ
2025	Charcot-Marie-Tooth (CMT)	Review	De Grado A, Serio M, Saveri P, Pisciotta C, Pareyson D. Charcot-Marie-Tooth disease: a review of clinical developments and its management - What's new in 2025? Expert	https://pubmed.ncbi.nlm.nih.gov/40014417/

			Rev Neurother. 2025 Apr;25(4):427-442. doi: 10.1080/14737175.2025.2470980. Epub 2025 Mar 7. PMID: 40014417.	
2024	Charcot-Marie-Tooth (CMT)	Review	McCray BA, Fridman V. Clinical Outcome Assessments and Biomarkers in Charcot-Marie-Tooth Disease. Neurology. 2024 Dec 24;103(12):e210120. doi: 10.1212/WNL.00000000000210120. Epub 2024 Nov 25. PMID: 39586049; PMCID: PMC11590233.	https://pubmed.ncbi.nlm.nih.gov/39586049/

Other NEUROMUSCULAR DISORDERS (2024-2025)

2025	Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP)	Review	Rajabally YA, Boggia GM, Riley D, Riley S, Peatman J, Noel W, Gary C, Nobile-Orazio E. Outcome measures in CIDP: A scoping and mapping review. J Neurol Sci. 2025 Oct 15;477:123654. doi: 10.1016/j.jns.2025.123654. Epub 2025 Aug 12. PMID: 40839897	https://pubmed.ncbi.nlm.nih.gov/40839897/
2025	Facioscapulohumeral Dystrophy (FSHD)	FSHD-RODS (PRO)	Teeselink S, Vincenten SCC, Voermans NC, van Alfen N, van Engelen BGM, Mul K. The Facioscapulohumeral Muscular Dystrophy Rasch-Built Overall Disability Scale (FSHD-RODS): Longitudinal Assessment of a Disease-Specific Patient Reported Outcome. Eur J Neurol. 2025 Aug;32(8):e70238. doi: 10.1111/ene.70238. PMID: 40781929; PMCID: PMC12334892.	https://pubmed.ncbi.nlm.nih.gov/40781929/
2024	Limb-Girdle Muscular Dystrophies (LGMD)	LGMD-HI (Health Index) (PRO)	Stouffer JA, Bates K, Thacker LR, Heatwole C, Johnson NE. The Limb Girdle Muscular Dystrophy Health Index (LGMD-HI). Neuromuscul Disord. 2024 Jun;39:48-53. doi: 10.1016/j.nmd.2024.04.008. Epub 2024 Apr 29. PMID: 38795602; PMCID: PMC11176010.	https://pubmed.ncbi.nlm.nih.gov/38795602/

2024	Limb-Girdle Muscular Dystrophies (LGMD)	Quantitative muscle MRI (qMRI)	Forsting J, Wächter M, Froeling M, Rohm M, Güttches AK, De Lorenzo A, Südkamp N, Kocabas A, Vorgerd M, Enax-Krumova E, Rehmann R, Schlaffke L. Quantitative muscle magnetic resonance imaging in limb-girdle muscular dystrophy type R1 (LGMDR1): A prospective longitudinal cohort study. NMR Biomed. 2024 Oct;37(10):e5172. doi: 10.1002/nbm.5172. Epub 2024 May 25. PMID: 38794994.	https://pubmed.ncbi.nlm.nih.gov/38794994/
2025	Myasthenia Gravis (NMJ)	PASS	Spagni G, Verza MU, Cornacchini S, Beretta F, Sun B, Lotti A, Falso S, Barilaro A, Masacesi L, Evoli A, Damato V. Validation of the "Patient-Acceptable Symptom State" Question as Outcome Measure in AChR Myasthenia Gravis: A Multicentre, Prospective Study. Eur J Neurol. 2025 Jun;32(6):e70262. doi: 10.1111/ene.70262. PMID: 40556475; PMCID: PMC12188101.	https://pubmed.ncbi.nlm.nih.gov/40556475/
2024	Myasthenia Gravis (NMJ)	MG-ADL (PRO)	Janssen MF, Dewilde S, Wolfe GI, Muppidi S, Phillips G. Psychometric properties of MG-ADL items and MG-ADL score: An assessment of distributional characteristics, validity and factor structure in two large datasets. J Neurol Sci. 2024 Aug 15;463:123135. doi: 10.1016/j.jns.2024.123135. Epub 2024 Jul 22. PMID: 39068745.	https://pubmed.ncbi.nlm.nih.gov/39068745/
2024	Spinal Muscular Atrophy (SMA)	HFMSE / RULM responsiveness (PerfO)	Hagenacker T, Maggi L, Coratti G, Youn B, Raynaud S, Paradis AD, Mercuri E. Effectiveness of Nusinersen in Adolescents and Adults with Spinal Muscular Atrophy: Systematic Review and Meta-analysis. Neurol Ther. 2024 Oct;13(5):1483-1504. doi: 10.1007/s40120-024-00653-2. Epub 2024 Sep 2. PMID: 39222296; PMCID: PMC11393259.	https://pubmed.ncbi.nlm.nih.gov/39222296/

We are most grateful for the generous support of this Conference by:



Co-funded by the
European Union

