

## Curriculum Vitae

**Luca Bello, MD, PhD**

### Work address:

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Luca Bello trained as a neurologist and neuromuscular clinician with Corrado Angelini and Elena Pegoraro at the University of Padova (UniPD). After completing residency (2012), he obtained a PhD in Neurosciences studying modifiers of dystrophinopathies with Elena Pegoraro and Eric P. Hoffman, spending 2 years at Children's National Research Institute in Washington, DC (2013-2014). As a faculty member at UniPD, he is continuing studies of natural history, treatment response, genetic modifiers, and genotype-phenotype correlations in dystrophinopathies, as well as in other neuromuscular disorders. He has collaborated extensively with several international consortia in these fields, such as the Cooperative International Neuromuscular Group (CINRG), the Italian DMD Network and Italian Association of Myology (AIM), the neuromuscular European Reference Network (ERN), the European NeuroMuscular Center (ENMC), TREAT-NMD, and the Jain Foundation. He teaches Neurology and Neurorehabilitation in several UniPD courses and is an Attending Neurologist at the Neurology Clinic of the University Hospital in Padova.

### Professional positions

- **May 2022 - present:** Associate Professor of Neurology, Department of Neurosciences, University of Padova, and Attending Neurologist, "Azienda Ospedale-Università Padova"
- **May 2019 - May 2022:** Assistant Professor (RTDb) of Neurology, Department of Neurosciences, University of Padova, and Attending Neurologist, "Azienda Ospedale-Università Padova"
- **February 2016 - January 2019:** Postdoctoral Fellow (Assegnista di Ricerca), Department of Neurosciences, University of Padova
- **July 2016 - March 2019:** Part-time Neurologist, IRCCS "San Camillo", Venice.
- **February 2013 - October 2014:** Postdoctoral Research Fellow, the Research Center for Genetic Medicine, Children's National Medical Center, Washington, DC (USA), in the laboratory of Eric P. Hoffman.
- **October 2012 - January 2013:** Part-time Neurologist, Neurology Clinic, "S. Antonio" Hospital, Padova.
- **August 2007 - July 2012:** Residency in Neurology, Neurology Clinic, University of Padova.
- **2007 - present:** Padova Board of Physicians, registration #10052

### Education

- **2013 - 2016:** PhD in Neuroscience, University of Padova. PhD thesis: "Molecular bases of clinical and phenotype variability in Duchenne and Becker muscular dystrophy" supervisors Prof. Elena Pegoraro, University of Padova, and Dr. Eric P. Hoffman, Children's National Medical Center, Washington, DC, USA.
- **2007 - 2012:** Residency in Neurology, Neurology Clinic, University of Padova.

- **2000 - 2006:** Degree in Medicine and Surgery, University of Padova (110/110 cum laude).
- **1992 - 2001:** Degree in Violin (9.50/10), Conservatory “Cesare Pollini”, Padova.
- **1994 - 2000:** High school degree (100/100 cum laude) at Liceo Ginnasio “Tito Livio”, Padova.

#### **Publications and bibliometric indexes (full list attached)**

- **111 original scientific papers on international peer-reviewed journals**, 13 as first author and 3 as last author;
- **10 review papers** and **2 book chapters**.
- **4122 citations, h-index 37** (source: Scopus ID 26649732700).

#### **Prizes, acknowledgements, fellowships, scientific societies, editorial activity**

- **2019:** indicated as a “rising star” in an international survey of Duchenne muscular dystrophy experts regarding key opinion leaders in the field, by Monocl.
- **2019-present:** Member of the European Association of Neurology Scientific Panel on muscle and neuromuscular junction disorders.
- **2018-2024:** Member of the elected Directive Council of the Italian Association of Myology, with functions of Treasurer.
- **2017:** “National Scientific Abilitation” certifying eligibility for the role of Associate Professor in Neurology (06/D6), recognized by the Italian Ministry of Research and University on March 28th, 2017.
- **2015, 2016, 2019:** Fellowships granted by the World Muscle Society for participation in the annual Meetings.
- **2016:** “Best publication” prize of the Italian Society of Neurology (SIN) for the paper “DMD genotypes and loss of ambulation in the CINRG Duchenne Natural History Study” (Bello et al., Neurology 2016).
- **2015:** “Best publication” prize of the Italian Society of Neurology (SIN) for the paper “Genetic modifiers of ambulation in the CINRG Duchenne Natural History Study” (Bello et al., Ann Neurol 2015).
- **2015:** “Best poster” prize of the Italian Association of Myology (AIM) for the paper “Genetic modifiers of ambulation in the CINRG Duchenne Natural History Study” (Bello et al., Ann Neurol 2015).
- **2014:** Selected as a Young Researcher participant for the 2014 Lindau Nobel Laureate Meetings for Physiology and Medicine, Lindau, Germany.
- **2010:** Fellowship granted by the Research Group on Neuromuscular Diseases-World Federation of Neurology for participation in the XII International Congress on Neuromuscular Diseases, Naples, Italy.
- **2000:** Ranked first out of 700+ competitive applicants to the University of Padova Medical School.
- Member of the Italian Society of Neurology, Italian Association of Myology, World Muscle society, and American Academy of Neurology.
- Peer review activity for Annals of Neurology, Neurology, Neuromuscular Disorders, Annals of Clinical and Translational Neurology, Chest, Acta Myologica, Journal of Neuromuscular Diseases, Scientific Reports, Muscle & Nerve, International Journal of Pediatrics, BioMed Research International, Neurological Sciences, Genes, Genomic Medicine, International Journal of Medical Sciences, and BMC Pediatrics.

### Specialization courses (attended as learner)

- **2015:** “Course Statistical Methods”, University of Padova.
- **2015:** “Complex Trait Analysis of Next Generation Sequence Data Course” at Max Delbrück Center for Molecular Medicine, Berlin, Germany.
- **2014:** Competitive selection for participation in the “Advanced Genome Mapping Course” sponsored by NIH at Rockefeller University, New York City, NY (USA).
- **2013:** “Proteomics” course at George Washington University, Integrated System Biology Program, Washington, DC, USA.

### Teaching and mentoring

<https://didattica.unipd.it/off/docente/D416558455E6CE88082D0B14B9958A2E>

- **2020-present:** Neurology course for Degree in Orthoptics, (20-hour course) at UniPD DNS (“Scienze Mediche” MEO2043848).
- **2019-present:** Neurology course for Degree in Auditory Prosthesis technicians (Treviso), 20-hour course at UniPD DNS (“Pediatria e Neurologia” MEO2044143).
- **2019-present:** Neurorehabilitation classes in the Masters course for rehabilitation (Laurea Magistrale in Scienze Riabilitative delle Professioni Sanitarie), 30-hour course, coordinator teacher of the overall 70-hour course, at UniPD DMM (“Scienze del Recupero” MEO2043403).
- **2019-present:** Member of the Council for the Neurology Residency school
- **2018-present:** Member of the Council for the PhD course in Translational Specialistic Medicine “G.B. Morgagni” at UniPD
- **2019-present:** Medical Statistics course for Residents in Neurology, 20-hour course, at UniPD.
- **2019-present:** Neuromuscular Medicine courses in the Genetics and Sports Medicine Residency schools at UniPD (10 hours each), at UniPD.
- **2020-present:** Responsible for the Quality Certification of teaching activities in the Neurology Residency school at UniPD.
- **2021-present:** coordinator of UniPD Medical Student rotations at the Neurology Clinic.
- Mentored and co-mentored 10 medical students, 5 Neurology residents, and 1 PhD candidate for their theses.

### Participation in clinical trials and observational studies

- **2024 – ongoing:** Efficacy, Safety and Tolerability of Givinostat in Non-ambulant Patients With Duchenne Muscular Dystrophy (ULYSSES, NCT05933057, Co-principal investigator).
- **2024 – ongoing:** Phase 2 Study of EDG-5506 in Becker Muscular Dystrophy (GRAND CANYON, NCT05291091, principal investigator).
- **2023 - ongoing:** A Study to Assess Vamorolone in Becker Muscular Dystrophy (NCT05166109, co-investigator)
- **2022 - 2023:** A Study of the Natural History of Participants With LGMD2E/R4, LGMD2D/R3, and LGMD2C/R5,  $\geq 4$  Years of Age, Who Are Managed in Routine Clinical Practice (NCT04475926, co-investigator).
- **2019 - 2020:** Study to Evaluate Amifampridine Phosphate in Patients With MuSK-MG (NCT03304054, co-investigator).
- **2017 - 2020:** A Phase III Double-blind Study With Idebenone in Patients With Duchenne Muscular Dystrophy (DMD) Taking Glucocorticoid Steroids (SIDEROS, NCT02814019, co-investigator).
- **2017 - ongoing:** Registry of Translarna (Ataluren) in Nonsense Mutation Duchenne Muscular Dystrophy (NCT02369731, co-investigator).

- **2013 - ongoing:** “Jain Foundation Clinical Outcome Study for Dyspherlinopathy” (NCT01676077, co-principal investigator).
- **2014 - 2020:** “Phase 3 trial for Finding the Optimum corticosteroid Regimen for Duchenne Muscular Dystrophy” (FOR-DMD, NCT01603407, co-investigator).
- **2014 - 2016:** “Phase 2-3 study of BYM338 in Sporadic Inclusion Body Myositis” (NCT02250443, co-investigator).
- **2014 - 2016:** “H6D-MC-LVJJ trial of Tadalafil for Duchenne muscular dystrophy” (NCT01865084, co-investigator).

#### **Organization of national and international conferences and workshops**

- **2024:** Organizer of the International Workshop “Optimize DMD” (Rome, November 2024)
- **2019-2024:** Faculty at the yearly National Congresses of the Italian Association of Myology
- **2022:** Organizer of the European Neuromuscular Center (ENMC) international Workshop: “Standards of care for the dysferlinopathies”

#### **Invited speaker at national and international conferences and workshops**

- **2019-2024:** Invited speaker at National Congresses of the Italian Association of Myology
- **2021 and 2024:** Invited speaker at World Muscle Society Conference (Industry Workshops)
- **2023:** Invited speaker at International Congress on Neuromuscular Diseases, digital edition

#### **Research Funding**

**2022-2025:** Telethon-UILDm Project GUP21003: “Characterizing Phenotypes in non ambulant Duchenne Muscular Dystrophy”. PI of Collaborating Center (Coordinator: Università Cattolica Sacro Cuore, Roma), €15,000.

**2023-2026:** Telethon-UILDm Project GUP23005: “Bone health in adult patients with Duchenne Muscular Dystrophy (DMD). A survey on osteoporosis, fragility fractures and bone-targeted therapies. PI of Collaborating Center (Coordinator: Istituto “Gaslini”, Genova), €25,000.

**2025-2028:** Telethon-UILDm Project GUP24005: “Respiratory progression in adult Duchenne muscular dystrophy. Natural history, identification of new biomarkers and design of a predictive algorithm”. PI of Collaborating Center (Coordinator: Istituto “Gaslini”, Genova), €46,750.

**2025-2027:** Telethon-UILDm Project GUP24011: “”. Co-PI of Collaborating Center (Coordinator: IRCCS “Medea”, Bosisio Parini), €17,000.

2025: Telethon Special Project: “Natural history in ambulant and non ambulant boys and young adults with Duchenne muscular dystrophy”. PI of Collaborating Center (Coordinator: Università Cattolica Sacro Cuore, Roma) GSP24002, €25,000

#### **Languages**

- First language: Italian.
- Excellent spoken and written English: Certificate of Proficiency in English, Cambridge, 1997, “A” degree; TOEFL iBT score 112/120, 98th %ile, 2007.

## Attachment: full list of publications

### Original peer reviewed scientific papers (starting from most recent)

1. Gorgoglione D, Sabbatini D, Riguzzi P, Capece G, Pane M, Servidei S, Briganti M, Sancricca C, Bruschi F, Ardisson A, Masson R, Gallone A, Maggi L, Picillo E, Politano L, Petrosino A, Vianello S, Penzo M, Villa M, Sframeli M, Allegra C, Barp A, Di Bari A, Salmin F, Albamonte E, Colacicco G, Panicucci C, Traverso M, Palermo C, Lerario A, Velardo D, D'Angelo MG, Berardinelli A, Gardani A, Nicotra R, Parravicini S, Siciliano G, Ricci G, Torri F, Gadaleta G, Urbano G, Rolle E, Ricci F, D'Amico A, Catteruccia M, Pini A, Giannotta M, Battini R, Marinella G, Previtali SC, Zambon AA, Ferlini A, Fortunato F, Magri F, Mongini TE, Sansone VA, Bruno C, Messina S, Nigro V, Moroni I, Mercuri E, **Bello L**, Pegoraro E. Natural history of Becker muscular dystrophy: DMD gene mutations predict clinical severity. *Brain*. 2024 Nov 5:awae358. doi: 10.1093/brain/awae358. Epub ahead of print. PMID: 39499670.
2. Salvalaggio A, Cacciavillani M, Tierro B, Coraci D, Currò R, Ferrarini M, Pegoraro E, **Bello L**, Fabrizi GM, Filla A, Padua L, Manganelli F, Cortese A, Briani C. Nerve ultrasound in CANVAS-spectrum disease: Reduced nerve size distinguishes genetically confirmed CANVAS from other axonal polyneuropathies. *J Peripher Nerv Syst*. 2024 Sep 2. doi: 10.1111/jns.12655. Epub ahead of print. PMID: 39219417. Borland H, Moore U, Dressman HG, Human A, Mayhew AG, Hilsden H, Rufibach LE, Duong T, Maron E, DeWolf B, Rose K, Siener C, Thiele S, Práxedes NS, Canal A, Holsten S, Sakamoto C, Pedrosa-Hernández I, Bello L, Alfano LN, Lowes LP The Jain COS Consortium, James MK, Straub V. Performance of upper limb entry item to predict forced vital capacity in dysferlin-deficient limb girdle muscular dystrophy. *Neuromuscul Disord*. 2024 Oct;43:20-28. doi: 10.1016/j.nmd.2024.08.003. Epub 2024 Aug 14. PMID: 39178649.
3. Govoni A, Ricci G, Bonanno S, **Bello L**, Magri F, Meneri M, Torri F, Caponnetto C, Passamano L, Grandis M, Trojsi F, Cerri F, Gadaleta G, Capece G, Caumo L, Tanel R, Saccani E, Vacchiano V, Sorarù G, D'Errico E, Tramacere I, Bortolani S, Rolle E, Gellera C, Zanin R, Silvestrini M, Politano L, Schenone A, Previtali SC, Berardinelli A, Turri M, Verriello L, Coccia M, Mantegazza R, Liguori R, Filosto M, Maioli MA, Simone IL, Mongini T, Corti S, Manca ML, Pegoraro E, Siciliano G, Comi GP, Maggi L. Six-minute walk test as outcome measure of fatigability in adults with spinal muscular atrophy treated with nusinersen. *Muscle Nerve*. 2024 Oct;70(4):816-823. doi: 10.1002/mus.28225. Epub 2024 Aug 2. PMID: 39096012.
4. Spagnoli C, Adorisio R, **Bello L**, D'Amico A, D'Angelo MG, Pane M, Penzo M, Riguzzi P, Sansone V, Vianello A, Fusco C. Continuity of care with ataluren in Duchenne Muscular Dystrophy patients with nonsense mutations after loss of ambulation. Personal experience. *Acta Myol*. 2023 Dec 31;42(4):118-122. doi: 10.36185/2532-1900-396. PMID: 38406379; PMCID: PMC10883323.
5. **Bello L**, Sabbatini D, Fusto A, Gorgoglione D, Borin GU, Penzo M, Riguzzi P, Villa M, Vianello S, Calore C, Melacini P, Vio R, Barp A, D'Angelo G, Gandossini S, Politano L, Berardinelli A, Messina S, Vita GL, Pedemonte M, Bruno C, Albamonte E, Sansone V, Baranello G, Masson R, Astrea G, D'Amico A, Bertini E, Pane M, Lucibello S, Mercuri E, Spurney C, Clemens P, Morgenroth L, Gordish-Dressman H, McDonald CM, Hoffman EP; CINRG-DNHS Investigators; Pegoraro E. The IAAM LTBP4 Haplotype is Protective Against Dystrophin-Deficient Cardiomyopathy. *J Neuromuscul Dis*. 2024;11(2):285-297. doi: 10.3233/JND-230129. PMID: 38363615.
6. Coratti G, Pane M, Brogna C, D'Amico A, Pegoraro E, **Bello L**, Sansone VA, Albamonte E, Ferraroli E, Mazzone ES, Fanelli L, Messina S, Sframeli M, Catteruccia M, Cicala G, Capasso A, Ricci M, Frosini S, De Luca G, Rolle E, De Sanctis R, Forcina N, Norcia G, Passamano L, Scutifero M, Gardani A, Pini A, Monaco G, D'Angelo MG, Leone D, Zanin R, Vita GL, Panicucci C, Bruno C, Mongini T, Ricci F, Berardinelli A, Battini R, Masson R, Baranello G, Dosi C, Bertini E, Nigro V, Politano L, Mercuri E. Gain and loss of upper limb abilities in Duchenne muscular dystrophy patients: A 24-month study. *Neuromuscul Disord*. 2024 Jan;34:75-82. doi: 10.1016/j.nmd.2023.11.011. Epub 2023 Dec 3. PMID: 38157655.
7. Musso G, Blasi L, Mion MM, Fortuna A, Sabbatini D, Zaninotto M, **Bello L**, Pegoraro E, Basso D, Plebani M, Sorarù G. Troponin T in spinal and bulbar muscular atrophy (SBMA). *J Neurol Sci*. 2024 Jan 15;456:122816. doi: 10.1016/j.jns.2023.122816. Epub 2023 Dec 1. PMID: 38071852.
8. Vicino A, **Bello L**, Bonanno S, Govoni A, Cerri F, Ferraro M, Capece G, Gadaleta G, Meneri M, Vacchiano V, Ricci G, D'Errico E, Tramacere I, Banfi P, Bortolani S, Zanin R, Maioli MA, Silvestrini M, Previtali SC, Berardinelli A, Turri M, Coccia M, Mantegazza R, Liguori R, Filosto M, Siciliano G, Simone IL, Mongini T, Comi G, Pegoraro E, Maggi L. Respiratory function in a large cohort of treatment-naïve adult spinal

- muscular atrophy patients: a cross-sectional study. *Neuromuscul Disord*. 2023 Dec;33(12):911-916. doi: 10.1016/j.nmd.2023.10.002. Epub 2023 Oct 12. PMID: 37945485.
9. Brogna C, Pane M, Coratti G, D'Amico A, Pegoraro E, **Bello L**, Sansone VAM, Albamonte E, Messina S, Pini A, D'Angelo MG, Bruno C, Mongini T, Ricci FS, Berardinelli A, Battini R, Masson R, Bertini ES, Politano L, Mercuri E, Italian Dmd Group. Upper Limb Changes in DMD Patients Amenable to Skipping Exons 44, 45, 51 and 53: A 24-Month Study. *Children (Basel)*. 2023 Apr 19;10(4):746. doi: 10.3390/children10040746. PMID: 37189996; PMCID: PMC10136754.
  10. Lupi A, Spolaor S, Favero A, **Bello L**, Stramare R, Pegoraro E, Nobile MS. Muscle magnetic resonance characterization of STIM1 tubular aggregate myopathy using unsupervised learning. *PLoS One*. 2023 May 8;18(5):e0285422. doi: 10.1371/journal.pone.0285422. PMID: 37155641; PMCID: PMC10166478.
  11. Pane M, Coratti G, Brogna C, Bovis F, D'Amico A, Pegoraro E, **Bello L**, Sansone V, Albamonte E, Ferraroli E, Mazzone ES, Fanelli L, Messina S, Catteruccia M, Cicala G, Ricci M, Frosini S, De Luca G, Rolle E, De Sanctis R, Forcina N, Norcia G, Passamano L, Gardani A, Pini A, Monaco G, D'Angelo MG, Capasso A, Leone D, Zanin R, Vita GL, Panicucci C, Bruno C, Mongini T, Ricci F, Berardinelli A, Battini R, Masson R, Baranello G, Dosi C, Bertini E, Politano L, Mercuri E. Longitudinal Analysis of PUL 2.0 Domains in Ambulant and Non-Ambulant Duchenne Muscular Dystrophy Patients: How do they Change in Relation to Functional Ability? *J Neuromuscul Dis*. 2023 Apr 12. doi: 10.3233/JND-221556. Epub ahead of print. PMID: 37066919.
  12. Canessa EH, Spathis R, Novak JS, Beedle A, Nagaraju K, **Bello L**, Pegoraro E, Hoffman EP, Hathout Y. Characterization of the dystrophin-associated protein complex by mass spectrometry. *Mass Spectrom Rev*. 2022 Nov 24:e21823. doi: 10.1002/mas.21823. Epub ahead of print. PMID: 36420714.
  13. Fortuna A, Sabbatini D, Frigo A, **Bello L**, Calvi F, Blasi L, Gianferrari G, Martinelli I, Minicuci G, Pegoraro E, Mandrioli J, Sorarù G. Italian version of the Rasch-Built Overall Amyotrophic Lateral Sclerosis Disability Scale (ROADS): validation and longitudinal performance. *J Neurol*. 2022 Nov 16. doi: 10.1007/s00415-022-11483-3. Epub ahead of print. PMID: 36383259.
  14. Maggi L\*, **Bello L\***, Bonanno S, Govoni A, Caponnetto C, Passamano L, Grandis M, Trojsi F, Cerri F, Gardani A, Ferraro M, Gadaleta G, Zangaro V, Caumo L, Maioli M, Tanel R, Saccani E, Meneri M, Vacchiano V, Ricci G, Sorarù G, D'Errico E, Bortolani S, Pavesi G, Gellera C, Zanin R, Corti S, Silvestrini M, Politano L, Schenone A, Previtali SC, Berardinelli A, Turri M, Verriello L, Coccia M, Mantegazza R, Liguori R, Filosto M, Marrosu G, Tiziano FD, Siciliano G, Simone IL, Mongini T, Comi G, Pegoraro E. Adults with spinal muscular atrophy: a large-scale natural history study shows gender effect on disease. *J Neurol Neurosurg Psychiatry*. 2022 Oct 11:jnnp-2022-329320. doi: 10.1136/jnnp-2022-329320. Epub ahead of print. PMID: 36220341.
  15. Kelley EF, Cross TJ, McDonald CM, Investigators C, Hoffman EP, Spurney CF, **Bello L**. Influence of  $\beta_1$  Adrenergic Receptor Genotype on Longitudinal Measures of Left Ventricular Ejection Fraction and Responsiveness to  $\beta$ -Blocker Therapy in Patients With Duchenne Muscular Dystrophy. *Clin Med Insights Cardiol*. 2022 Aug 23;16:11795468221116838. doi: 10.1177/11795468221116838. PMID: 36046180; PMCID: PMC9421016.
  16. Gregorio I, Mereu M, Contarini G, **Bello L**, Semplicini C, Burgio F, Russo L, Sut S, Dall'Acqua S, Braghetta P, Semenza C, Pegoraro E, Papaleo F, Bonaldo P, Cescon M. Collagen VI deficiency causes behavioral abnormalities and cortical dopaminergic dysfunction. *Dis Model Mech*. 2022 Sep 1;15(9):dmm049481. doi: 10.1242/dmm.049481. Epub 2022 Sep 21. PMID: 35946603; PMCID: PMC9548377.
  17. Abbonante V, Malara A, Chrisam M, Metti S, Soprano P, Semplicini C, **Bello L**, Bozzi V, Battiston M, Pecci A, Pegoraro E, De Marco L, Braghetta P, Bonaldo P, Balduini A. Lack of COL6/collagen VI causes megakaryocyte dysfunction by impairing autophagy and inducing apoptosis. *Autophagy*. 2022 Jul 20:1-16. doi: 10.1080/15548627.2022.2100105. Epub ahead of print. PMID: 35857791.
  18. Coratti G, Lenkowicz J, Norcia G, Lucibello S, Ferraroli E, d'Amico A, **Bello L**, Pegoraro E, Messina S, Ricci F, Mongini T, Berardinelli A, Masson R, Previtali SC, D'angelo G, Magri F, Comi GP, Politano L, Passamano L, Vita G, Sansone VA, Albamonte E, Panicucci C, Bruno C, Pini A, Bertini E, Patarnello S, Pane M, Mercuri E; italian DMD study group. Age, corticosteroid treatment and site of mutations affect motor functional changes in young boys with Duchenne Muscular Dystrophy. *PLoS One*. 2022 Jul 29;17(7):e0271681. doi: 10.1371/journal.pone.0271681. PMID: 35905042; PMCID: PMC9337636.q
  19. Schiava M, Ikenaga C, Villar-Quiles RN, Caballero-Ávila M, Topf A, Nishino I, Kimonis V, Udd B, Schoser B, Zanolati E, Souza PVS, Tasca G, Lloyd T, Lopez-de Munain A, Paradas C, Pegoraro E, Nadaj-Pakleza A, De Bleecker J, Badrising U, Alonso-Jiménez A, Kostera-Pruszczyk A, Miralles F, Shin JH, Bevilacqua JA, Olivé M, Vorgerd M, Kley R, Brady S, Williams T, Domínguez-González C, Papadimas GK, Warman-Chardon J, Claeys KG, de Visser M, Muelas N, LaForet P, Malfatti E, Alfano LN, Nair SS, Manousakis G, Kushlaf HA, Harms MB,

- Nance C, Ramos-Fransi A, Rodolico C, Hewamadduma C, Cetin H, García-García J, Pál E, Farrugia ME, Lamont PJ, Quinn C, Nedkova-Hristova V, Peric S, Luo S, Oldfors A, Taylor K, Ralston S, Stojkovic T, Wehl C, Diaz-Manera J; VCP International Study Group; VCP International Study Group. Genotype-phenotype correlations in valosin-containing protein disease: a retrospective multicentre study. *J Neurol Neurosurg Psychiatry*. 2022 Jul 27:jnnp-2022-328921. doi: 10.1136/jnnp-2022-328921. Epub ahead of print. PMID: 35896379.
20. Sabbatini D, Fusto A, Vianello S, Villa M, Janik J, D'Angelo G, Diella E, Magri F, Comi GP, Panicucci C, Bruno C, D'Amico A, Bertini E, Astrea G, Battini R, Politano L, Masson R, Baranello G, Previtali SC, Messina S, Vita G, Berardinelli A, Mongini T, Pini A, Pane M, Mercuri E, Hoffman EP, Morgenroth L, Gordish-Dressman H, Duong T, McDonald CM, **Bello L**, Pegoraro E. Genetic modifiers of upper limb function in Duchenne muscular dystrophy. *J Neurol*. 2022 May 5. doi: 10.1007/s00415-022-11133-8. Epub ahead of print. PMID: 35513612.
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