CURRICULUM VITAE
Dr Federica Trucco, MD, PhD

Personal Information:

Name (including title): Dr Federica Trucco, MD, PhD
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Professional registration Medici e Chirurghi di Genova, OMGE 15346
GMC Number: 7565579 Full specialist registration
English Level C1 (test IELTS Academic 8.0/9), Portuguese and Spanish Good Level

Affiliations

- Dept of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Care (DINOGMI), University of Genova, Paediatric Neurology and Muscle Disease Unit, IRCCS Istituto 'Giannina Gaslini', Genova, Italy
- Honorary fellow at Dubowitz Neuromuscular Centre, GOSH-ICH, UCL, London, UK
- Honorary consultant at Paediatric Respiratory Department, Royal Brompton Hospital, London, UK

Best education and training

- Nov 2016 – May 2020 – PhD in Paediatrics, Curriculum Metabolic, Neurodegenerative, Neuromuscular paediatric disorders – University of Genova (Main Supervisor Prof. C. Minetti)

- July 2014 Specialization in Paediatrics (University of Genova, G. Gaslini Institute)

Best positions

- November 2023 – to date Senior researcher (Ricercatore tipo B a tempo determinato) in Paediatric Neurology Dept of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Care (DINOGMI), University of Genova, Genova, Italy

- April 2022 – October 2023 International Fellowship at Neurorehabilitation Unit, University of Milan - Nemo Clinical Center, Milan

- Jan 2022 – to date Honorary contract Dubowitz Neuromuscular Centre – Great Ormond Street Hospital- Institute of Child Health, University College London, London, UK
Honorary Consultant in Paediatric Sleep Medicine – Royal Brompton Hospital, London, UK

- Jan 2020 – Jan 2022 Consultant in Paediatric Sleep Medicine – Joint post Evelina Children Hospital (Guys and St Thomas) – Royal Brompton Hospital. Honorary contract Dubowitz Neuromuscular Centre – Great Ormond Street Hospital-Institute of Child Health, University College London

- April 2018 – December 2019 Neuromuscular Clinical Research Fellow at Dubowitz Neuromuscular Centre – Great Ormond Street Hospital-Institute of Child Health, University College London

- Jan 2017 – Mar 2018 PhD student and Honorary Clinical Research Fellow (Partecipating), Paediatrics Respiratory Department, Royal Brompton Hospital

- Sep 2014 – Dec 2016 Neuromuscular Clinical Research Fellow in Neuromuscular Unit in G.

Gaslini Pediatric Institute, Supervisor Prof. C. Minetti

Awards

2017-2018 European Respiratory Society Funded Short Term Research Fellowship
Project "Respiratory muscle strength tests as predictors of hypoventilation in neuromuscular children"

Oct 2019 Elsevier Runner-up Prize poster – 24th Congress World Muscle Society
"Respiratory function in SMA type 2 and non-ambulant SMA type 3 longitudinal data from the international SMA consortium (iSMAc)"

2016, 2019 Education Fund Fellowship award - World Muscle Society Conference

Jan 2020 Award American Thoracic society - Assembly on Pediatrics 2020 scholarship
"Inflammation in Children with Neuromuscular Disorders and Sleep Disordered Breathing"

Grants

Sep 2014 Sub-Investigator "Multicentric pilot project of home tele-monitoring in paediatric patients requiring mechanical ventilation'' awarded by Italian Ministry of Health to Prof. C. Minetti

Feb 2018 Principal Investigator "Respiratory muscle strength tests as predictors of hypoventilation in neuromuscular children" awarded by European Respiratory Society

Jan 2024 Principal Investigator "Sleep features of Myotonic Dystrophy type 1" awarded by Research funds of Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Genova, Italy

March 2024 Principal Investigator "Identification of predictors of respiratory failure in patients with Duchenne Muscular Dystrophy'' awarded by Parent Project association

August 2024 Principal Investigator "Respiratory progression in adult Duchenne muscular dystrophy. Natural history, identification of new biomarkers and design of a predictive algorithm" awarded by Fondazione Telethon-UILDM

Invited presentations at national and international conferences

Conference Sleep disorders in childhood, Genova 19-21 June 2024, *"Sleep disorders in children with neuromuscular disorders"*

Workshop SMArt Rising together, Milan 18/06/2024 *"Unmet needs in patients with SMA"*

23rd Conference Italian Association of Myology June 2024, Symposium *"The new generation of SMA patients"*

Workshop Precision Medicine in Duchenne Muscular Dystrophy, 11-12 November 2023 *"The clinical approach to patients with Duchenne Muscular Dystrophy"*

Workshop Infants 0-12 Months with Spinal Muscular Atrophy, Milano, 26/05/2023 - *"La fisiologia del neonato e infante sano"*

Workshop SMAkers 2023, 2024 - *"Tavola Rotonda: focus sulla gestione del paziente pediatrico" and "Workshop bulbar function in SMA"*

SMA international Preceptorship, Milano, 25-26/11/2022 - *"Respiratory Management of patients with SMA"*

22nd Conference Italian Association of Myology 2022- Symposium *"Nuove traiettorie di patologia nella SMA – come deve evolvere l'approccio multidisciplinare"*

2nd Sleep and Ventilation, Pediatric Meeting (Jornadas De Sueño Y Ventilación En Pediatría) organizzata dalla Società Argentina di Sonno, Sessione ibrida, 29-30/09/2022 - *"Respiratory PROS and CONS of the new treatments for SMA, DMD and other neuromuscular diseases"*

14th European Paediatric Neurology Society 2022, Glasgow, 28/04/2022-02/05/2022 Simposio *"More than movement:other meaningful milestones in Type 1 SMA"*

SMArt Network Webinar Series (Novartis Taiwan), 21/04/2022 – *"Pulmonary management of the new phenotype of Spinal muscular atrophy"*

Treat-NMD European Expert Masterclass on Congenital Myopathies 2022 – *"Respiratoryinvolvement in paediatric congenital myopathies"*

International Conference Neuromuscular disorders (ICNMD) 2021 *"Cardiorespiratoryfunction in Duchenne Muscular Dystrophy"*

Italian SMAcademy conference 2021 *"Respiratory natural history in Spinal MuscularAtrophy"*

British Myology Society UK Annual conference 2020 *"Effects of corticosteroids on cardiorespiratory function in DMD"*

Italian Spinal Muscular Atrophy Association Annual Conference 2019 *"Respiratory function in patients with SMA type 2 and SMA type 3 non ambulant"*

Authorship of books' chapter

- Invited author of the section "Limb girdle muscular dystrophies" for the chapter "Neuromuscular disorders" in the book "Neurologia Pediatrica" edited by Prof. Martino Ruggieri, Issued by Edra in August 2022 · EAN: 9788821448652 · ISBN: 8821448657.

- Invited author of the chapter "Pulmonary pathophysiology of neuromuscular disease" for the book published by the American Thoracic Society "Respiratory Assessment and management of Paediatric Neuromuscular Disease" edited by Dr. Oscar Henry Mayer issued on 13th April 2023 by Elsevier. Paperback ISBN: 9780323957472.eBook ISBN: 9780323957489.

Involvement in scientific networks, steering committees, advisory boards

- Invited member of UK NorthStar network Respiratory Working group and coauthor of the *"Respiratory care Guidelines for Duchenne Muscular Dystrophy"* endorsed by British Thoracic Society and accepted for publication in Thorax, October 2023.
- Invited speaker at the international meeting *"Respiratory Assessment and Management of Duchenne Muscular Dystrophy: Current Standards and Future Directions"* promoted by Parent Project association Italia and US.
- Invited speaker to the international **283th ENMC workshop** 'Establishing Expert Care Recommendations for LAMA2-RDs: A Prototype for the Development of Congenital Muscular Dystrophy Subtype-Specific Care Guidelines'
- Invited speaker to the international **284th ENMC workshop** 'Cognitive and behavioral abnormalities in pediatric DM1 – what should we measure in preparation for clinical trials?'

- Invited member of International Spinal Muscular Atrophy consortium (iSMAc) and Italian network for Duchenne Muscular Dystrophy.
- Invited member of the *"International Scientific Steering committee to develop recommendations about the clinical management of the key aspect of the X-Linked Myotubular Myopathy (XLMTM)"* promoted by Astellas therapeutics.
- Selected to participate to the workshop *"Foundation for the future in Myotonic Dystrophy: Developing leaders to tackle challenges in Myotonic Dystrophy"* promoted by the RNA Institute, University of Albany, Washington – 20-22/09/2023
- Invited member of the Advisory Scientific Working Group meeting for Roche *"SMA Digital Assessment tools"* held online and in Zurich on 9/05/2023.

Sub-Investigator in clinical trials for

- Duchenne muscular dystrophy (Exon skipping of exon 45, 51, Sarepta; Givinostat, Italfarmaco; Vamorolone, ReveraGen BioPharma; Pamrevlumab, Fibrogen)
- Spinal muscular atrophy type 1 (Ionis 39664-CS3B and CS11 with intrathecal Nusinersen, Biogen; Gene therapy with Onasemnogene abeparvovec, Novartis) and type 2 (Jewelfish trial with Risdiplam, Roche)
- Myotubular myopathy (Inceptus , Audentes therapeutics)

- Review Editor for Frontiers in Neurology – specialty section Neuromuscular Disorders and Peripheral Neuropathies since September 2022.

- Organizer and chair of the first *"Anita Simonds symposium – Turning the tide, Respiratory management of neuromuscular disorders in the era of new treatments"*, held virtually on the 28th May 2021
- Organiser and chair of the symposium *"New era of disease modifying treatments in childhood neuromuscular disorders: Changing landscapes in sleep diagnostics and management"* selected at the International Paediatric Sleep Association conference 2024 in Glasgow
- Chair of the section *"AIM giovani"* part of the Italian association of myology (AIM)

Membership

- Member of the Italian Association of Myology
- Member of the British myology society
- Member of the British sleep society
- Member of the Italian Society of Respiratory Medicine

Relevant publications

Author of 45 manuscripts published in peer-review journals

H index 17 Scopus; H index 18 Google scholar

1. **Trucco F**, O'Halloran KD. *Unravelling the complexity of respiratory involvement in Duchenne muscular dystrophy: An urgent call for a collective translational approach*. **J Physiol.** 2025 May 31.
2. Bello L, Riguzzi P, Albamonte E, Astrea G, Battini R, Barp A, Berardinelli AL, Bertini ES, Brolatti N, Bruno C, Corti S, D'Amico A, D'Angelo MG, Dallavalle G, Liguori R, Maggi L, Magri F, Mancuso M, Masson R, Mercuri E, Minetti C, Messina S, Mongini T, Musumeci O, Nigro V, Pane M, Panicucci C, Pedemonte M, Pini A, Politano L, Previtali S, Ricci F, Ricci G, Ruggiero L, Sansone V, Siciliano G, Trabacca A, **Trucco F**, Velardo D, Pegoraro E, Comi GP. *Opinion of the Italian Association of Myology on Ataluren for the Treatment of Nonsense Mutation Duchenne Muscular Dystrophy.* **Drugs R D.** 2025 May 28.

3. Racca F, Finder JD, Mercuri E, Vianello A, Chatwin M, Amaddeo A, Crescimanno G, **Trucco F**, Sheehan DW, Barberis L, Buccella F, Wolff J, Katz SL, Birnkrant DJ; PPMD Respiratory Group. *Current Standards and Future Directions of Duchenne Muscular Dystrophy Respiratory Care: The PPMD Italy Meeting Report.* **Pediatr Pulmonol.** 2025 May;60(5):e71113.

4. Capasso A, Cicala G, Ricci M, Pane M, D'Amico A, Bruno C, Sansone VA, Messina S, Bello L, Pegoraro E, D'Angelo MG, Masson R, Berardinelli A, Pini A, Ricci F, Mongini TE, Coccia M, Nigro V, Trabacca A, Filosto M, Comi G, Magri F, Barp A, Battini R, Previtali SC, Valentino ML, Diella E, Dosi C, Ruggiero L, Siciliano G, Ricci G, Catteruccia M, Arpaia C, Coratti G, Norcia G, Bonanno S, Verriello L, Agosto C, Varone A, Ferlini A, Maioli MA, Brogna C, Siliquini S, Bruno I, Panicucci C, Allegra C, Albamonte E, Mercuri E; Italian DMD group. *Prevalence of Duchenne muscular dystrophy in Italy: a nationwide survey.* **Eur J Pediatr.** 2024 Dec 16;184(1):86. doi: 10.1007/s00431-024-05903-x.

5. **Trucco F**, Lizio A, Roma E, Di Bari A, Salmin F, Albamonte E, Casiraghi J, Pozzi S, Becchiati S, Antonaci L, Salvalaggio A, Catteruccia M, Tosi M, Marinella G, Danti F.R., Bruschi F, Veneruso M, Parravicini S, Fiorillo C, Berardinelli A, Pini A, Moroni I, Astrea G, Battini R, D'Amico A, Ricci F, Pane M, Mercuri E.M, Johnson N.E, Sansone V.A. *Association between Reported Sleep Disorders and Behavioral Issues in Children with Myotonic Dystrophy Type 1—Results from a Retrospective Analysis in Italy.* **J Clin Med.** 2024 Sep 14;13(18):5459. doi: 10.3390/jcm13185459.

6. **Trucco F**, Albamonte E, Pane M, Ricci F, D'amico A, Astrea G, Moroni I, Pini A, Fiorillo C, Berardinelli A, Johnson NE, Sansone VA; DM1 PAEDIATRIC WORKING GROUP. *Parental diagnostic delay and developmental outcomes in congenital and childhood-onset myotonic dystrophy type 1.* **Dev Med Child Neurol.** 2024 Sep 4. doi: 10.1111/dmcn.16079. Online ahead of print.

7. Santangelo A, Corsello A, Gizzi G, Lancieri M, Diana MC, **Trucco F,** Orsini A, Bonuccelli A, Peroni DG, Perilli L, Correnti E, Santangelo G, Striano P, Raieli V. *Exploring Headaches in Pediatric Behçet Disease: Prevalence, Clinical Impact, and Management.* **J Clin Med.** 2024 Jun 23;13(13):3659.

8. **Trucco F**, Ridout D, Weststrate H, Scoto M, Rohwer A, Coratti G, Main M, Mayhew A, Montes J, De Sanctis R, Pane M, Pera MC, Sansone VA, Albamonte E, D'Amico A, Bruno C, Messina S, Childs AM, Willis T, Ong M, Servais L, Majumdar A, Hughes I, Marini-Bettolo C, Parasuraman D, Gowda V, Baranello G, Bertini E, De Vivo DC, Darras BT, Day JW, Mayer OH, Zolkipli-Cunningham Z, Finkel RS, Mercuri E, Muntoni F, on behalf of the iSMAC group. *Therapeutic role of Nusinersen on respiratory progression in paediatric patients with spinal muscular atrophy type 2 and non-ambulant type 3*. **Neurol Clin Pract.** 2024 Jun;14(3):e200298.

9. **Trucco F.,** Davies M., Zambon A.A., Ridout D., Abel F., Muntoni F. *Definition of diaphragmatic sleep disordered breathing and clinical meaning in Duchenne Muscular Dystrophy.* **Thorax.** 2024 May 9:thorax-2023-220729.

10. McGrattan K, Cerchiari A, Conway E, Berti B, Finkel R, Muntoni F, Mercuri E; **iSMAc working group.** *Bulbar function in spinal muscular atrophy (SMA): state of art and future challenges. 21 July 2023, Rome, Italy.* **Neuromuscul Disord.** 2024 May;38:44-50.

11. **Trucco F**, Dastagir S, Tan HL. Pseudo-obstructive sleep disordered breathing – definition and progression in Spinal Muscular Atrophy. **Sleep Med.** 2024 Mar;115:61-65

12. N. Brolatti*, **F. Trucco***, M. Ferretti*, C. Avanti, P. Tacchetti, C. Panicucci, P. Striano, C. Minetti, C. Bruno, M. Pedemonte. Structured Light Plethysmography for Non-invasive assessment of respiratory pattern in Spinal Muscular Atrophy type 1. **J Clin Med. 2023 Dec 7;12(24):7553.**

13. Childs AM, Turner C, Astin R, Bianchi S, Cunningham V, Edel L, Edwards C, Farrant P, Heraghty J, James M, Massey C, Messer B, Michel-Sodhi J, Murphy P, Schiava M, Thomas A, **Trucco F**, Guglieri M. *Development of respiratory care guidelines for Duchenne muscular dystrophy in the UK: key recommendations for clinical practice. Consensus of the DMD Care UK Respiratory Working Group*. **Thorax 2023. Dec 20:thorax-2023-220811. Online ahead of print.**

14. **Trucco F.**, Salmin F., Lizio A., Coratti G., Albamonte E., Frisoni M.C., Mauro L., Carraro E., Palazzo G., Lops J., Cattaneo C., Pozzi S., Casiraghi J., Di Bari A., Berti B., Stanca G., Ricci M., Pane M., Heatwole C., Dilek N., Mercuri

E., Sansone V.A. *Assessing prevalence and characteristics of oro-bulbar involvement in children and adults with SMA type 2 and 3 using a multimodal approach.* **Dysphagia. 2023** Jun 8.

15. Mercuri E, Seferian AM, Servais L, Deconinck N, Stevenson H, Ni X, Zhang W, East L, Yonren S, Muntoni F; **4658-102 Study Group:** Nicolas Deconinck, Rudy Van Coster, Arnaud Vanlander, Andreea Seferian, Silvana De Lucia, Teresa Gidaro, Laura Vanden Brande, Laurent Servais, Janbernd Kirschner, Sabine Borell, Eugenio Mercuri, Claudia Brogna, Marika Pane, Lavinia Fanelli, Giulia Norcia, Francesco Muntoni, Chiara Brusa, Mary Chesshyre, Kate Maresh, Jaqueline Pitchforth, Lucia Schottlaender, Mariacristina Scoto, Arpana Silwal, **Federica Trucco**. *Safety, tolerability and pharmacokinetics of eteplirsen in young boys aged 6-48 months with Duchenne muscular dystrophy amenable to exon 51 skipping.* **Neuromuscul Disord. 2023** Mar 24;33(6):476-483.

16. Chiriboga CA, Bruno C, Duong T, Fischer D, Mercuri E, Kirschner J, Kostera-Pruszczyk A, Jaber B, Gorni K, Kletzl H, Carruthers I, Martin C, Warren F, Scalco RS, Wagner KR, Muntoni F; **JEWELFISH Study Group.** *Risdiplam in Patients Previously Treated with Other Therapies for Spinal Muscular Atrophy: An Interim Analysis from the JEWELFISH Study.* **Neurol Ther. 2023** Apr;12(2):543-557.

17. Croci C, Cataldi M, Baratto S, Bruno C, **Trucco F**, Doccini S, Romano A, Nesti C, Santorelli FM, Fiorillo C. *Recurrent Sensory-Motor Neuropathy Mimicking CIDP as Predominant Presentation of PDH Deficiency.* **Neuropediatrics. 2023** Jun;54(3):211-216. doi: 10.1055/a-2018-4845. Epub 2023 Jan 24.

18. Zambon AA, **Trucco F**, Laverty A, Riley M, Ridout M, Manzur AY, Abel F, and Muntoni F.*Respiratory function and sleep disordered breathing in pediatric Duchenne Muscular Dystrophy.* **Neurology. 2022** Sep 20;99(12):e1216-e1226

19. **Trucco F.** *Daytime predictors of nocturnal hypercapnic hypoventilation in children with neuromuscular disorders-The Holy Grail.* **Pediatr Pulmonol. 2022** Jun;57(6):1377-1379. doi:10.1002/ppul.25888.

20. Zambon AA, Gupta VA, Ridout D, Manzur AY, Baranello G, **Trucco F**, Muntoni F on behalf ofUK NorthStar Clinical Network. *Peak functional ability is associated with age at loss of ambulation in Duchenne Muscular Dystrophy*. **Dev Med Child Neurol.** 2022 Aug;64(8):979- 988. doi: 10.1111/dmcn.15176

21. **Trucco F**, Ridout D, Domingos J, Maresh K, Chesshyre M, Munot P, Sarkozy A, Robb S, Quinlivan R, Riley M, Wallis C, Chan E, Abel F, De Lucia S, Hogrel JY, Niks EH, de Groot I, Servais L, Straub V, Ricotti V, Manzur A, Muntoni F; UK NorthStar Clinical Network and AFMNetwork. *Genotype-related respiratory progression in Duchenne Muscular Dystrophy - multicentre international study.* **Muscle Nerve**. 2021 Oct 4. doi: 10.1002/mus.27427. Onlineahead of print.

22. Jones S, Hanwell R, Chowdhury T, Orgill J, van den Eshof K, Farquhar M, Joseph D, Gringras Pand **Trucco F.** *Feasibility and parental perception of home sleep studies during COVID-19: a tertiary sleep centre experience.* **Arch Dis Child**. 2021 Sep 22: archdischild-2021-322184.

23. **Trucco F**, Ridout D, Scoto M, Coratti G, Main ML, Lofra RM, Mayhew AG, Montes J, Pane M, Sansone V, Albamonte E, D'Amico A, Bertini E, Messina S, Bruno C, Parasuraman D, Childs AM, Gowda V, Willis T, Ong M, Marini-Bettolo C, De Vivo DC, Darras BT, Day J, Kichula EA, Mayer OH, Navas Nazario AA, Finkel RS, Mercuri E, Muntoni F; international SMA consortium(iSMAc). *Respiratory trajectories in type 2 and non-ambulant 3 Spinal muscular atrophy in the iSMAC cohort study.* **Neurology.** 2020 Oct 16:10.1212.

24. **Trucco F**, Domingos J, Tay CG, Ridout D, Maresh K, Munot P, Sarkozy A, Robb S, Quinlivan Ros, Riley M, Burch M, Fenton M, Wallis C, Chan E, Abel F, Manzur AY, Muntoni F. *Long-termcardiorespiratory progression and role of corticosteroids in Duchenne muscular dystrophy.* **Chest.** 2020 Oct;158(4):1606-1616. doi: 10.1016/j.chest.2020.04.043. Epub 2020 May 7.

25. Wadman RI, De Amicis R, Brusa C, Battezzati A, Bertoli S, Davis T, Main M, Manzur A, Mastella C, Munot P, Imbrigiotta N, Schottlaender L, Sarkozy A, **Trucco F**, Baranello G, ScotoM, Muntoni F. *Feeding difficulties in children and adolescents with spinal muscular atrophy type 2.* **Neuromuscul Disord.** 2021 Feb;31(2):101-112. doi: 10.1016/j.nmd.2020.12.007. Epub 2020 Dec 19.

26. **Trucco F,** Carruthers E, Davies JC, Simonds A, Bush A, Tan HL. *Inflammation in children with neuromuscular disorders and sleep disordered breathing.* **Sleep Med.** 2020 Aug;72:118-121. doi: 10.1016/j.sleep.2020.03.032. Epub 2020 Apr 9.

27. Bello L, D'Angelo G, Villa M, Fusto A, Vianello S, Merlo B, Sabbatini D, Barp A, Gandossini S,Magri F, Comi GP, Pedemonte M, Tacchetti P, Lanzillotta V, **Trucco F**, D'Amico A, Bertini E, Astrea G, Politano L, Masson R, Baranello G, Albamonte E, De Mattia E, Rao F, Sansone VA, Previtali S, Messina S, Vita GL, Berardinelli A, Mongini T, Pini A, Pane M, Mercuri E, VianelloA, Bruno C, Hoffman EP, Morgenroth L, Gordish-Dressman H, McDonald CM; CINRG-DNHS Investigators, Pegoraro E. *Genetic modifiers of respiratory function in Duchenne muscular dystrophy.* **Ann Clin Transl Neurol.** 2020 Apr 28.

28. **Trucco F,** Rosenthal M, Bush A, Tan HL. *McGill score as a screening test for obstructive sleepdisordered breathing in children with co-morbidities.* **Sleep Med.** 2020 Apr;68:173-176. doi: 10.1016/j.sleep.2019.12.010. Epub 2019 Dec 28.

29. Fiorillo C, D'Apice MR, **Trucco F**, Murdocca M, Spitalieri P, Assereto S, Baratto S, Morcaldi G,Minetti C, Sangiuolo F, Novelli G. *Characterization of MDPL Fibroblasts Carrying the Recurrent p.Ser605del Mutation in POLD1 Gene.* **DNA Cell Biol.** 2018 Nov 2.

30. **Trucco F,** Bush A, Tan HL. *Reducing the need for carbon dioxide monitoring in the investigation of paediatric sleep disordered breathing.* **Eur Respir J.** 2018 Oct 4;52(4). pii:1801290.

31. **Trucco F,** Chatwin M, Semple T, Rosenthal M, Bush A, Tan HL. *Sleep disordered breathingand ventilatory support in children with Down syndrome.* **Pediatr Pulmonol.** 2018 Oct;53(10):1414-1421.

32. **Trucco F,** Pedemonte M, Racca F, Falsaperla R, Romano C, Wenzel A, D'Agostino A, PistorioA, Tacchetti P, Bella C, Bruno C, Minetti C. *Tele-monitoring in paediatric and young home- ventilated neuromuscular patients: A multicentre case-control trial.* **J Telemed Telecare.** 2018 Jan 1:1357633X18778479.

33. **Trucco F,** Pedemonte M, Fiorillo C, Tan HL, Carlucci A, Brisca G, Tacchetti P, Bruno C, Minetti C. *Detection of early nocturnal hypoventilation in neuromuscular disorders.* **J Int Med Res.** 2018 Mar;46(3):1153-1161.

34. Finkel RS, Mercuri E, Darras BT, Connolly AM, Kuntz NL, Kirschner J, Chiriboga CA, Saito K, Servais L, Tizzano E, Topaloglu H, Tulinius M, Montes J, Glanzman AM, Bishop K, Zhong ZJ,Gheuens S, Bennett CF, Schneider E, Farwell W, De Vivo DC; **ENDEAR Study Group.** Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. N Engl J Med. 2017 Nov 2;377(18):1723-1732.

35. **Trucco F**, Pedemonte M, Fiorillo C, Tacchetti P, Brisca G, Bruno C, Minetti C. Respiratorypattern in a FSHD pediatric population. **Respir Med.** 2016 Oct;119:78-80. doi: 10.1016/j.rmed.2016.08.014. Epub 2016 Aug 22.

36. Panicucci C, Fiorillo C, Moro F, Astrea G, Brisca G, **Trucco F**, Pedemonte M, Lanteri P, Sciarretta L, Minetti C, Santorelli FM, Bruno C. *Mutations in GMPPB Presenting with Pseudometabolic Myopathy.* **JIMD Rep.** 2018;38:23-31. doi: 10.1007/8904_2017_25. Epub2017 Apr 30.

37. D'Amico A, Catteruccia M, Baranello G, Politano L, Govoni A, Previtali SC, Pane M, D'Angelo MG, Bruno C, Messina S, Ricci F, Pegoraro E, Pini A, Berardinelli A, Gorni K, Battini R, Vita G, **Trucco F,** Scutifero M, Petillo R, D'Ambrosio P, Ardissone A, Pasanisi B, Vita G, Mongini T, Moggio M, Comi GP, Mercuri E, Bertini E. *Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements.* **Neuromuscul Disord.** *2017 May;27(5):447-451. doi: 10.1016/j.nmd.2017.02.006. Epub 2017 Feb 14.*

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39. Fiorillo C, Astrea G, Savarese M, Cassandrini D, Brisca G, **Trucco F**, Pedemonte M, Trovato R, Ruggiero L, Vercelli L, D'Amico A, Tasca G, Pane M, Fanin M, Bello L, Broda P, Musumeci O, Rodolico C, Messina S, Vita GL, Sframeli M, Gibertini S, Morandi L, Mora M, Maggi L, PetrucciA, Massa R, Grandis M, Toscano A, Pegoraro E, Mercuri E, Bertini E, Mongini T, Santoro L, Nigro V, Minetti C, Santorelli FM, Bruno C; Italian Network on Congenital Myopathies.
MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. **Orphanet J Rare Dis.** 2016 Jul 7;11(1):91. doi: 10.1186/s13023-016-0476-1.

40. Fiorillo C, Moro F, Brisca G, Accogli A, **Trucco F**, Trovato R, Pedemonte M, Severino M, CatalaM, Capra V, Santorelli FM, Bruno C, Rossi A, Minetti C. *Beyond spinal muscular atrophy with lower extremity dominance: cerebellar hypoplasia associated with a novel mutation in BICD2.***Eur J Neurol.** 2016 Apr;23(4):e19-21. doi: 10.1111/ene.12914.

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