

CURRICULUM VITAE

NOME E COGNOME	NICOLA TICOZZI
LUOGO E DATA DI NASCITA	MILANO, 25 FEBBRAIO 1979
NAZIONALITÀ	ITALIANA
CODICE FISCALE	TCZNCL79B25F205C
INDIRIZZO	VIA PATTI 4, MILANO
TELEFONO	02-61911-2937
FAX	02-61911-2038
EMAIL	N.TICOZZI@AUXOLOGICO.IT NICOLA.TICOZZI@UNIMI.IT

ISTRUZIONE E FORMAZIONE

- 2017: Dottorato di Ricerca in Neuroscienze, Rudolph Magnus Brain Center, Utrecht University
- 2009: Specializzazione in Neurologia (70/70 e lode), Università degli Studi di Milano
- 2008-2009: Research Fellow presso i Dipartimenti di Neurologia del Massachusetts General Hospital, Harvard Medical School (Charlestown, MA, USA) e della University of Massachusetts Medical School (Worcester, MA, USA)
- 2004: Laurea in Medicina e Chirurgia (110/110 e lode), Università degli Studi di Milano

POSIZIONE ATTUALE

- Professore Associato (MED/26 - Neurologia) presso il Dipartimento di Fisiopatologia Medico-Chirurgica e dei Trapianti, Università degli Studi di Milano, con funzioni assistenziali presso l'U.O. Neurologia, Istituto Auxologico Italiano, IRCCS (2019-presente)

POSIZIONI PRECEDENTI

- 2016-2019: Ricercatore a Tempo Determinato tipo B (MED/26 - Neurologia), Dipartimento di Fisiopatologia Medico-Chirurgica e dei Trapianti, Università degli Studi di Milano
- 2012-2015: Ricercatore a Tempo Determinato, tipo A (MED/26 - Neurologia), Dipartimento di Fisiopatologia Medico-Chirurgica e dei Trapianti, Università degli Studi di Milano
- 2009-2012 e 2015-2016: Neurologo Ricercatore, U.O. Neurologia, Istituto Auxologico Italiano, IRCCS

ABILITAZIONI E CERTIFICAZIONI

- Abilitazione all'esercizio della professione di Medico e Chirurgo nel novembre 2004
- Iscritto all'Albo dei Medici e Chirurghi di Milano dal 01.03.2005 (n°39422)
- Fellow dell'European Board of Neurology (2013)
- Abilitazione a Professore di I fascia (MED/26 – Neurologia) conseguita il 26/04/2021 (validità 9 anni)

SKETCH BIOGRAFICO

Nato a Milano il 25 febbraio 1979, il Prof. Ticozzi si è laureato cum laude presso la Facoltà di Medicina e Chirurgia dell'Università degli Studi di Milano nel 2004, dove ha anche completato la sua specializzazione in Neurologia nel 2009 sotto la guida del Prof. Vincenzo Silani. E' stato Research Fellow presso il Day Laboratory for Neuromuscular Research, Massachusetts General Hospital e presso il Dipartimento di Neurologia, University of Massachusetts Medical School (2008-2009).

Attualmente Nicola Ticozzi è Professore Associato di Neurologia presso l'Università degli Studi di Milano, e Neurologo Ricercatore presso il Dipartimento di Neurologia, Istituto Auxologico Italiano IRCCS. In questa posizione, ha acquisito una vasta conoscenza nella diagnosi e nella cura dei pazienti con sclerosi laterale amiotrofica (SLA) e altre malattie neurodegenerative, ha partecipato all'organizzazione di studi clinici e organizzato il database clinico e la biobanca dell'Istituto. La sua principale area di ricerca è la genetica delle malattie del motoneurone e di altre malattie neurodegenerative. In particolare, ha studiato un'ampia coorte di pazienti SLA per mutazioni patogenetiche in geni causativi, contribuendo così alla comprensione dell'epidemiologia genetica della malattia in Italia. È stato anche responsabile della raccolta dei dati clinici per il Consorzio SLAGEN, che ha condotto un vasto studio GWAS sulla SLA sporadica. Infine, ha co-fondato una partnership internazionale volta a identificare nuovi fattori di rischio genetici nella SLA: questa collaborazione ha portato alla scoperta di diversi nuovi geni malattia, tra cui *PFN1*, *TUBA4A*, *TBK1*, *NEK1*, *ANXA11*, *CCNF* e *KIF5A*.

Il Prof. Ticozzi è co-autore di 94 articoli peer-reviewed, e, in qualità di esperto di genetica della SLA, è stato invitato come revisore per diverse riviste scientifiche e come relatore a diversi convegni nazionali e internazionali (SIN, EAN, EFNS). È membro del Comitato Clinico-scientifico di AISLA - Associazione Italiana SLA, e Fellow dell'European Board of Neurology. È stato P.I. di numerosi progetti di ricerca, e ha ricevuto finanziamenti da enti pubblici e privati senza scopo di lucro.

ATTIVITÀ SCIENTIFICA

Indici di produzione scientifica

- Co-autore di 94 pubblicazioni in riviste indicizzate
- Co-autore di 6 capitoli di libri
- Editor di 1 libro
- H-index: 32
- Citazioni: 6136

Società scientifiche

- Società Italiana di Neurologia
- European Academy of Neurology

Attività editoriale

- Associate Editor di *Frontiers in Aging Neuroscience*
- Guest Editor di *International Journal of Molecular Sciences*
- Revisore per numerose riviste scientifiche tra cui *Human Molecular Genetics*, *Human Mutation*, *Journal of Neurology*, *Journal of Neurology Neurosurgery and Psychiatry*, *PLoS One*, *BMJ Open*, *Amyotrophic Lateral Sclerosis and Frontotemporal Dementia* e *Neurological Sciences*.

Revisore scientifico per agenzie di finanziamento alla ricerca

- *French National Research Agency, AFM-Telethon, National Science Center - NCN Poland, Stichting Alzheimer Onderzoek/Fondation Recherche Alzheimer - SAO/FRA*

LETTURE MAGISTRALI E ORGANIZZAZIONE DI CONGRESSI SCIENTIFICI

Relatore invitato a >20 congressi nazionali e internazionali, tra cui:

- *"Controversies in MND: genetics of ALS"*, 51° Congresso SIN, Novembre 2020, Milano.
- *"Clinical trials in ALS"*, 48° Congresso SIN, Ottobre 2017, Napoli.
- *"RNA-related neurological disorders and RNA interference in therapy: ALS"*, 1° Congresso EAN, Giugno 2015, Berlino.
- *"Genetics of ALS/FTD"*, 45° Congresso SIN, Ottobre 2014, Cagliari.
- *"Recent updates on ALS"*, 40° Congresso SIN, Novembre 2009, Padova.
- *"Protein aggregation and defective RNA metabolism as mechanisms for motor neuron damage"*, 13° Congresso EFNS, Settembre 2009, Pisa.

Organizzazione di congressi scientifici:

- Membro del comitato organizzatore locale del 51° Congresso SIN, Novembre 2020, Milano (virtuale).
- Membro del comitato organizzatore del 14° Congresso ENCALS, Maggio 2016, Milano.
- Membro del comitato organizzatore di >10 congressi scientifici e corsi di aggiornamento ospitati dall'Istituto Auxologico Italiano, IRCCS.

FINANZIAMENTI DA PROGETTI COMPETITIVI *PEER-REVIEWED*

- 2021: P.I. del progetto *"Gender-ALS - Gender modulation of C9orf72 toxicity in amyotrophic lateral sclerosis"*. Università degli Studi di Milano, Bando SEED 2019.
- 2021: P.I. del progetto *"AZYGOS 2.0 - Autozygosity mapping followed by next generation sequencing in unrelated consanguineous individuals to identify novel ALS-associated genes"*. AriSLA - Fondazione Italiana di Ricerca per la SLA.
- 2018: P.I. del progetto *"GR-2016-02364373 - DRepALS - The role of the novel NEK1 and c21orf2 genes associated to DNA repair in the pathogenesis of amyotrophic lateral sclerosis"*. Ministero della Salute, Bando Giovani Ricercatori - Ricerca Finalizzata 2016.
- 2014: P.I. del progetto *"GR-2011-02347820 - IRisALS - Identification of Risk variants associated with susceptibility to Sporadic Amyotrophic Lateral Sclerosis using an exome sequencing approach"*. Ministero della Salute, Bando Giovani Ricercatori - Ricerca Finalizzata 2011-2012.
- 2013: Coordinatore scientifico del progetto *"NOVALS - Identification of de novo mutations in amyotrophic lateral sclerosis using a trio-based exome sequencing approach"*. AriSLA - Fondazione Italiana di Ricerca per la SLA.
- 2010: Coordinatore scientifico del progetto *"EXOMEFALS - Identification of candidate disease genes in FALS using a targeted exome capture and resequencing approach"*. AriSLA - Fondazione Italiana di Ricerca per la SLA.

ATTIVITÀ DIDATTICA

- Responsabile del Corso Integrato di Infermieristica Clinica della Disabilità Neuropsichica, Corso di Laurea in Infermieristica (sezioni San Giuseppe e Fondazione Don Gnocchi), Università degli Studi di Milano
- Docente nell'ambito del corso di Clinica delle Malattie Neurologiche, Corso di Laurea in Medicina e Chirurgia, Università degli Studi di Milano
- Docente nell'ambito della Scuola di Specializzazione in Neurologia, Università degli Studi di Milano
- Docente nell'ambito della Scuola di Dottorato di Medicina Traslazionale (A.A. 2015-2016), Università degli Studi di Milano

ELENCO DELLE PUBBLICAZIONI PEER-REVIEWED

1. Poletti B, Solca F, Carelli L, Diena A, Colombo E, Torre S, Maranzano A, Greco L, Cozza F, Lizio A, Ferrucci R, Girotti F, Verde F, Morelli C, Lunetta C, Silani V, Ticozzi N. Association of Clinically Evident Eye Movement Abnormalities With Motor and Cognitive Features in Patients With Motor Neuron Disorders. *Neurology*. 2021 Sep 9;10.1212/WNL.0000000000012774. doi: 10.1212/WNL.0000000000012774. Epub ahead of print. PMID: 34504031.
2. Johnson JO, Chia R, Miller DE, Li R, Kumaran R, Abramzon Y, Alahmady N, Renton AE, Topp SD, Gibbs JR, Cookson MR, Sabir MS, Dalgard CL, Troakes C, Jones AR, Shatunov A, Iacoangeli A, Al Khleifat A, Ticozzi N, Silani V, Gellera C, Blair IP, Dobson-Stone C, Kwok JB, Bonkowski ES, Palvadeau R, Tienari PJ, Morrison KE, Shaw PJ, Al-Chalabi A, Brown RH Jr, Calvo A, Mora G, Al-Saif H, Gotkine M, Leigh F, Chang IJ, Perlman SJ, Glass I, Scott AI, Shaw CE, Basak AN, Landers JE, Chiò A, Crawford TO, Smith BN, Traynor BJ; FALS Sequencing Consortium; American Genome Center; International ALS Genomics Consortium; and ITALSGEN Consortium, Smith BN, Ticozzi N, Fallini C, Gkazi AS, Topp SD, Scotter EL, Kenna KP, Keagle P, Tiloca C, Vance C, Troakes C, Colombrita C, King A, Pensato V, Castellotti B, Baas F, Ten Asbroek ALMA, McKenna-Yasek D, McLaughlin RL, Polak M, Asress S, Esteban-Pérez J, Stevic Z, D'Alfonso S, Mazzini L, Comi GP, Del Bo R, Ceroni M, Gagliardi S, Querin G, Bertolin C, van Rheenen W, Rademakers R, van Blitterswijk M, Lauria G, Duga S, Corti S, Cereda C, Corrado L, Sorarù G, Williams KL, Nicholson GA, Blair IP, Leblond-Manry C, Rouleau GA, Hardiman O, Morrison KE, Veldink JH, van den Berg LH, Al-Chalabi A, Pall H, Shaw PJ, Turner MR, Talbot K, Taroni F, García-Redondo A, Wu Z, Glass JD, Gellera C, Ratti A, Brown RH Jr, Silani V, Shaw CE, Landers JE, Dalgard CL, Adeleye A, Soltis AR, Alba C, Viollet C, Bacikova D, Hupalo DN, Sukumar G, Pollard HB, Wilkerson MD, Martinez EM, Abramzon Y, Ahmed S, Arepalli S, Baloh RH, Bowser R, Brady CB, Brice A, Broach J, Campbell RH, Camu W, Chia R, Cooper-Knock J, Ding J, Drepper C, Drory VE, Dunckley TL, Eicher JD, England BK, Faghri F, Feldman E, Floeter MK, Fratta P, Geiger JT, Gerhard G, Gibbs JR, Gibson SB, Glass JD, Hardy J, Harms MB, Heiman-Patterson TD, Hernandez DG, Jansson L, Kirby J, Kowall NW, Laaksovirta H, Landeck N, Landi F, Le Ber I, Lumbroso S, MacGowan DJL, Maragakis NJ, Mora G, Mouzat K, Murphy NA, Myllykangas L, Nalls MA, Orrell RW, Ostrow LW, Pamphlett R, Pickering-Brown S, Pioro EP, Pletnikova O, Pliner HA, Pulst SM, Ravits JM, Renton AE, Rivera A, Robberecht W, Rogaeva E, Rollinson S, Rothstein JD, Scholz SW, Sendtner M, Shaw PJ, Sidle KC, Simmons Z, Singleton AB, Smith N, Stone DJ, Tienari PJ, Troncoso JC, Valori M, Van Damme P, Van Deerlin VM, Van Den Bosch L, Zinman L, Landers JE, Chiò A, Traynor BJ, Angelocola SM, Ausiello FP, Barberis M, Bartolomei I, Battistini S, Bersano E, Bisogni G, Borghero G, Brunetti M, Cabona C, Calvo A, Canale F, Canosa A, Cantisani TA, Capasso M, Caponnetto C, Cardinali P, Carrera P, Casale F, Chiò A, Colletti T, Conforti FL, Conte A, Conti E, Corbo M, Cuccu S, Dalla Bella E, D'Errico E, DeMarco G, Dubbioso R, Ferrarese C, Ferraro PM, Filippi M, Fini N, Floris G, Fuda G, Gallone S, Gianferrari G, Giannini F, Grassano M, Greco L, Iazzolino B, Introna A, La Bella V, Lattante S, Lauria G, Liguori R, Logroscino G, Logullo FO, Lunetta C, Mandich P, Mandrioli J, Manera U, Manganelli F, Marangi G, Marinou K, Marrosu MG, Martinelli I, Messina S, Moglia C, Mora G, Mosca L, Murru MR, Origone P, Passaniti C, Petrelli C, Petrucci A, Pozzi S, Pugliatti M, Quattrini A, Ricci C, Riolo G, Riva N, Russo M, Sabatelli M, Salamone P, Salivetto M, Salvi F, Santarelli M, Sbaiz L, Sideri R, Simone I, Simonini C, Spataro R, Tanel R, Tedeschi G, Ticca A, Torriello A, Tranquilli S, Tremolizzo L, Trojsi F, Vasta R, Vacchiano V, Vita G, Volanti P, Zollino M, Zucchi E. Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurol*. 2021 Aug 30:e212598. doi: 10.1001/jamaneurol.2021.2598. Epub ahead of print. PMID: 34459874; PMCID: PMC8406220.
3. Greco LC, Lizio A, Casiraghi J, Sansone VA, Tremolizzo L, Riva N, Solca F, Torre S, Ticozzi N, Filippi M, Silani V, Poletti B, Lunetta C. A preliminary comparison between ECAS and ALS-CBS in classifying cognitive-behavioural phenotypes in a cohort of non-demented amyotrophic lateral sclerosis patients. *J Neurol*. 2021 Aug 19. doi: 10.1007/s00415-021-10753-w. Epub ahead of print. PMID: 34410493.
4. Tagini S, Brugnera A, Ferrucci R, Mazzocco K, Pievani L, Priori A, Ticozzi N, Compare A, Silani V, Pravettoni G, Poletti B. Attachment, Personality and Locus of Control: Psychological Determinants of Risk Perception and Preventive Behaviors for COVID-19. *Front Psychol*. 2021 Jul 9;12:634012. doi: 10.3389/fpsyg.2021.634012. PMID: 34305708; PMCID: PMC8299752.

5. Colombo E, Messina S, Verde F, Locatelli M, Poletti B, Silani V, Ticozzi N. Epileptic Capgras-Like Delusions in a Patient with Right Frontal Meningioma: Case Report. *Case Rep Neurol.* 2021 May 27;13(2):284-288. doi: 10.1159/000513675. PMID: 34177534; PMCID: PMC8215999.
6. Pasquini J, Maremmani C, Salvadori S, Silani V, Ticozzi N. Testing olfactory dysfunction in acute and recovered COVID-19 patients: a single center study in Italy. *Neurol Sci.* 2021 Jun;42(6):2183-2189. doi: 10.1007/s10072-021-05200-7. Epub 2021 Mar 26. PMID: 33768438; PMCID: PMC7994059.
7. Silani V, Corcia P, Harms MB, Rouleau G, Siddique T, Ticozzi N. Genetics of primary lateral sclerosis. *Amyotroph Lateral Scler Frontotemporal Degener.* 2020 Nov;21(sup1):28-34. doi: 10.1080/21678421.2020.1837177. PMID: 33602012.
8. Shephard SR, Parker MD, Cooper-Knock J, Verber NS, Tuddenham L, Heath P, Beauchamp N, Place E, Sollars ESA, Turner MR, Malaspina A, Fratta P, Hewamadduma C, Jenkins TM, McDermott CJ, Wang D, Kirby J, Shaw PJ; Project MINE Consortium; Project MinE. Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. *J Neurol Neurosurg Psychiatry.* 2021 May;92(5):510-518. doi: 10.1136/jnnp-2020-325014. Epub 2021 Feb 14. PMID: 33589474; PMCID: PMC8053339.
9. Cozza F, Lizio A, Greco LC, Bona S, Donvito G, Carraro E, Tavazzi S, Ticozzi N, Poletti B, Sansone VA, Lunetta C. Ocular Involvement Occurs Frequently at All Stages of Amyotrophic Lateral Sclerosis: Preliminary Experience in a Large Italian Cohort. *J Clin Neurol.* 2021 Jan;17(1):96-105. doi: 10.3988/jcn.2021.17.1.96. PMID: 33480204; PMCID: PMC7840315.
10. Moisse M, Zwamborn RAJ, van Vugt J, van der Spek R, van Rheenen W, Kenna B, Van Eijk K, Kenna K, Corcia P, Couratier P, Vourc'h P, Hardiman O, McLaughlin R, Gotkine M, Drory V, Ticozzi N, Silani V, de Carvalho M, Mora Pardina JS, Povedano M, Andersen PM, Weber M, Başak NA, Chen X, Eberle MA, Al-Chalabi A, Shaw C, Shaw PJ, Morrison KE, Landers JE, Glass JD, Robberecht W, van Es M, van den Berg L, Veldink J, Van Damme P; Project MinE Sequencing Consortium. The Effect of SMN Gene Dosage on ALS Risk and Disease Severity. *Ann Neurol.* 2021 Apr;89(4):686-697. doi: 10.1002/ana.26009. Epub 2021 Jan 15. PMID: 33389754; PMCID: PMC8048961.
11. Cooper-Knock J, Zhang S, Kenna KP, Moll T, Franklin JP, Allen S, Nezhad HG, Iacoangeli A, Yacovzada NY, Eitan C, Hornstein E, Elhaik E, Celadova P, Bose D, Farhan S, Fishilevich S, Lancet D, Morrison KE, Shaw CE, Al-Chalabi A; Project MinE ALS Sequencing Consortium, Veldink JH, Kirby J, Snyder MP, Shaw PJ. Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. *Cell Rep.* 2020 Dec 1;33(9):108456. doi: 10.1016/j.celrep.2020.108456. Erratum in: *Cell Rep.* 2021 Feb 2;34(5):108730. PMID: 33264630; PMCID: PMC7710676.
12. Dewan R, Chia R, Ding J, Hickman RA, Stein TD, Abramzon Y, Ahmed S, Sabir MS, Portley MK, Tucci A, Ibáñez K, Shankaracharya FNU, Keagle P, Rossi G, Caroppo P, Tagliavini F, Waldo ML, Johansson PM, Nilsson CF; American Genome Center (TAGC); FALS Sequencing Consortium; Genomics England Research Consortium; International ALS/FTD Genomics Consortium (iAFGC); International FTD Genetics Consortium (IFGC); International LBD Genomics Consortium (iLBDGC); NYGC ALS Consortium; PROSPECT Consortium, Rowe JB, Benussi L, Binetti G, Ghidoni R, Jabbari E, Viollet C, Glass JD, Singleton AB, Silani V, Ross OA, Ryten M, Torkamani A, Tanaka T, Ferrucci L, Resnick SM, Pickering-Brown S, Brady CB, Kowal N, Hardy JA, Van Deerlin V, Vonsattel JP, Harms MB, Morris HR, Ferrari R, Landers JE, Chiò A, Gibbs JR, Dalgard CL, Scholz SW, Traynor BJ. Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Neuron.* 2021 Feb 3;109(3):448-460.e4. doi: 10.1016/j.neuron.2020.11.005. Epub 2020 Nov 26. PMID: 33242422; PMCID: PMC7864894.
13. Scialò C, Tran TH, Salzano G, Novi G, Caponnetto C, Chiò A, Calvo A, Canosa A, Moda F, Caroppo P, Silani V, Ticozzi N, Ratti A, Borroni B, Benussi L, Ghidoni R, Furlanis G, Manganotti P, Senigaglia B, Parisse P, Bresselet R, Buratti E, Legname G. TDP-43 real-time quaking induced conversion reaction optimization and detection of seeding activity in CSF of amyotrophic lateral sclerosis and frontotemporal dementia patients. *Brain Commun.* 2020 Sep 14;2(2):fcaa142. doi: 10.1093/braincomms/fcaa142. PMID: 33094285; PMCID: PMC7566418.
14. Verde F, Zaina G, Bodio C, Borghi MO, Soranna D, Peverelli S, Ticozzi N, Morelli C, Doretti A, Messina S, Maderna L, Colombrita C, Gumina V, Tiloca C, Meroni PL, Zambon A, Ratti A, Silani V. Cerebrospinal fluid phosphorylated neurofilament heavy chain and chitotriosidase in primary lateral sclerosis. *J Neurol Neurosurg Psychiatry.* 2021 Feb;92(2):221-223. doi: 10.1136/jnnp-2020-324059. Epub 2020 Aug 27. PMID: 32855294.
15. Schito P, Ceccardi G, Calvo A, Falzone YM, Moglia C, Lunetta C, Marinou K, Ticozzi N, Scialò C, Sorarù G, Trojsi F, Conte A, Tortelli R, Russo M, Zucchi E, Pozzi L, Domi T, Carrera P, Agosta F, Quattrini A, Fazio R, Chiò A, Sansone VA, Mora G, Silani V, Volanti P, Caponnetto C, Querin G, Tedeschi G, Sabatelli M, Logroscino G, Messina S, Mandrioli J, Riva N, Filippi M. Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. *J Neurol Neurosurg Psychiatry.* 2020 Sep;91(9):1001-1003. doi: 10.1136/jnnp-2020-323542. Epub 2020 Jul 10. PMID: 32651246.
16. Carlomagno C, Banfi PI, Gualerzi A, Picciolini S, Volpato E, Meloni M, Lax A, Colombo E, Ticozzi N, Verde F, Silani V, Bedoni M. Human salivary Raman fingerprint as biomarker for the diagnosis of Amyotrophic Lateral Sclerosis. *Sci Rep.* 2020 Jun 23;10(1):10175. doi: 10.1038/s41598-020-67138-8. PMID: 32576912; PMCID: PMC7311476.
17. Poletti B, Carelli L, Lunetta C, Ticozzi N, Silani V. Advance care planning and mental capacity in ALS: a current challenge for an unsolved matter. *Neurol Sci.* 2020 Oct;41(10):2997-2998. doi: 10.1007/s10072-020-04462-x. Epub 2020 May 20. PMID: 32435995.

18. Schijven D, Stevelink R, McCormack M, van Rheenen W, Luykx JJ, Koeleman BPC, Veldink JH; Project MinE ALS GWAS Consortium; International League Against Epilepsy Consortium on Complex Epilepsies. Analysis of shared common genetic risk between amyotrophic lateral sclerosis and epilepsy. *Neurobiol Aging*. 2020 Aug;92:153.e1-153.e5. doi: 10.1016/j.neurobiolaging.2020.04.011. Epub 2020 Apr 18. PMID: 32409253; PMCID: PMC7818383.
19. Pensato V, Magri S, Bella ED, Tannorella P, Bersano E, Sorarù G, Gatti M, Ticozzi N, Taroni F, Lauria G, Mariotti C, Gellerà C. Sorting Rare ALS Genetic Variants by Targeted Re-Sequencing Panel in Italian Patients: *OPTN*, *VCP*, and *SQSTM1* Variants Account for 3% of Rare Genetic Forms. *J Clin Med*. 2020 Feb 3;9(2):412. doi: 10.3390/jcm9020412. PMID: 32028661; PMCID: PMC7073901.
20. Morelli C, Tiloca C, Colombrita C, Zambon A, Soranna D, Lafronza A, Solca F, Carelli L, Poletti B, Doretti A, Verde F, Maderna L, Ticozzi N, Ratti A, Silani V. CSF angiogenin levels in amyotrophic lateral Sclerosis-Frontotemporal dementia spectrum. *Amyotroph Lateral Scler Frontotemporal Degener*. 2020 Feb;21(1-2):63-69. doi: 10.1080/21678421.2019.1704016. Epub 2019 Dec 18. PMID: 31852251.
21. Trojsi F, Siciliano M, Femiano C, Santangelo G, Lunetta C, Calvo A, Moglia C, Marinou K, Ticozzi N, Ferro C, Scialò C, Sorarù G, Conte A, Falzone YM, Tortelli R, Russo M, Sansone VA, Chiò A, Mora G, Silani V, Volanti P, Caponnetto C, Querin G, Sabatelli M, Riva N, Logroscino G, Messina S, Fasano A, Monsurrò MR, Tedeschi G, Mandrioli J. Comparative Analysis of *C9orf72* and Sporadic Disease in a Large Multicenter ALS Population: The Effect of Male Sex on Survival of *C9orf72* Positive Patients. *Front Neurosci*. 2019 May 17;13:485. doi: 10.3389/fnins.2019.00485. PMID: 31156370; PMCID: PMC6534038.
22. Zucchi E, Ticozzi N, Mandrioli J. Psychiatric Symptoms in Amyotrophic Lateral Sclerosis: Beyond a Motor Neuron Disorder. *Front Neurosci*. 2019 Mar 11;13:175. doi: 10.3389/fnins.2019.00175. PMID: 30914912; PMCID: PMC6421303.
23. Verde F, Tiloca C, Morelli C, Doretti A, Poletti B, Maderna L, Messina S, Gentilini D, Fogh I, Ratti A, Silani V, Ticozzi N. *PON1* is a disease modifier gene in amyotrophic lateral sclerosis: association of the Q192R polymorphism with bulbar onset and reduced survival. *Neurol Sci*. 2019 Jul;40(7):1469-1473. doi: 10.1007/s10072-019-03834-2. Epub 2019 Mar 22. PMID: 30903418.
24. Poletti B, Carelli L, Solca F, Pezzati R, Faini A, Ticozzi N, Mitsumoto H, Silani V. Sexuality and intimacy in ALS: systematic literature review and future perspectives. *J Neurol Neurosurg Psychiatry*. 2019 Jun;90(6):712-719. doi: 10.1136/jnnp-2018-319684. Epub 2018 Dec 11. PMID: 30538137.
25. Tiloca C, Sorosina M, Esposito F, Peroni S, Colombrita C, Ticozzi N, Ratti A, Martinelli-Boneschi F, Silani V. Response to the commentary "The effect of *C9orf72* intermediate repeat expansions in neurodegenerative and autoimmune diseases" by Biasiotto G and Zanella I.^{*}. *Mult Scler Relat Disord*. 2019 Jan;27:79-80. doi: 10.1016/j.msard.2018.10.007. Epub 2018 Oct 14. PMID: 30347338.
26. Tiloca C, Sorosina M, Esposito F, Peroni S, Colombrita C, Ticozzi N, Ratti A, Martinelli-Boneschi F, Silani V. No *C9orf72* repeat expansion in patients with primary progressive multiple sclerosis. *Mult Scler Relat Disord*. 2018 Oct;25:192-195. doi: 10.1016/j.msard.2018.07.047. Epub 2018 Aug 1. PMID: 30099204.
27. Poletti B, Carelli L, Faini A, Solca F, Meriggi P, Lafronza A, Ciringione L, Pedroli E, Ticozzi N, Ciammola A, Cipresso P, Riva G, Silani V. The Arrows and Colors Cognitive Test (ACCT): A new verbal-motor free cognitive measure for executive functions in ALS. *PLoS One*. 2018 Aug 9;13(8):e0200953. doi: 10.1371/journal.pone.0200953. PMID: 30091987; PMCID: PMC6084851.
28. de Majo M, Topp SD, Smith BN, Nishimura AL, Chen HJ, Gkazi AS, Miller J, Wong CH, Vance C, Baas F, Ten Asbroek ALMA, Kenna KP, Ticozzi N, Redondo AG, Esteban-Pérez J, Tiloca C, Verde F, Duga S, Morrison KE, Shaw PJ, Kirby J, Turner MR, Talbot K, Hardiman O, Glass JD, de Bellerocche J, Gellerà C, Ratti A, Al-Chalabi A, Brown RH, Silani V, Landers JE, Shaw CE. ALS-associated missense and nonsense *TBK1* mutations can both cause loss of kinase function. *Neurobiol Aging*. 2018 Nov;71:266.e1-266.e10. doi: 10.1016/j.neurobiolaging.2018.06.015. Epub 2018 Jun 25. PMID: 30033073; PMCID: PMC6983933.
29. Poletti B, Solca F, Carelli L, Faini A, Madotto F, Lafronza A, Monti A, Zago S, Ciammola A, Ratti A, Ticozzi N, Abrahams S, Silani V. Cognitive-behavioral longitudinal assessment in ALS: the Italian Edinburgh Cognitive and Behavioral ALS screen (ECAS). *Amyotroph Lateral Scler Frontotemporal Degener*. 2018 Aug;19(5-6):387-395. doi: 10.1080/21678421.2018.1473443. Epub 2018 May 26. PMID: 29804470.
30. Carelli L, Solca F, Faini A, Madotto F, Lafronza A, Monti A, Zago S, Doretti A, Ciammola A, Ticozzi N, Silani V, Poletti B. The Complex Interplay Between Depression/Anxiety and Executive Functioning: Insights From the ECAS in a Large ALS Population. *Front Psychol*. 2018 Apr 5;9:450. doi: 10.3389/fpsyg.2018.00450. PMID: 29674987; PMCID: PMC5895754.
31. Nicolas A, Kenna KP, Renton AE, Ticozzi N, Faghri F, Chia R, Dominov JA, Kenna BJ, Nalls MA, Keagle P, Rivera AM, van Rheenen W, Murphy NA, van Vugt JJFA, Geiger JT, Van der Spek RA, Pliner HA, Shankaracharya, Smith BN, Marangi G, Topp SD, Abramzon Y, Gkazi AS, Eicher JD, Kenna A; ITALSGEN Consortium, Mora G, Calvo A, Mazzini L, Riva N, Mandrioli J, Caponnetto C, Battistini S, Volanti P, La Bella V, Conforti FL, Borghero G, Messina S, Simone IL, Trojsi F, Salvi F, Logullo FO, D'Alfonso S, Corrado L, Capasso M, Ferrucci L; Genomic Translation for ALS Care (GTAC) Consortium, Moreno CAM, Kamalakaran S, Goldstein DB; ALS Sequencing Consortium, Gitler AD, Harris T, Myers RM; NYGC ALS Consortium, Phatnani H, Musunuri RL, Evani US, Abhyankar A, Zody MC; Answer ALS Foundation, Kaye J, Finkbeiner S, Wyman SK, LeNail A, Lima L, Fraenkel E, Svendsen CN, Thompson LM, Van Eyk JE, Berry JD, Miller TM, Kolb SJ, Cudkowicz M, Baxi

- E; Clinical Research in ALS and Related Disorders for Therapeutic Development (CReATe) Consortium, Benatar M, Taylor JP, Rampersaud E, Wu G, Wu J; SLAGEN Consortium, Lauria G, Verde F, Fogh I, Tiloca C, Comi GP, Sorarù G, Cereda C; French ALS Consortium, Corcia P, Laaksovirta H, Myllykangas L, Jansson L, Valori M, Ealing J, Hamdalla H, Rollinson S, Pickering-Brown S, Orrell RW, Sidle KC, Malaspina A, Hardy J, Singleton AB, Johnson JO, Arepalli S, Sapp PC, McKenna-Yasek D, Polak M, Asress S, Al-Sarraj S, King A, Troakes C, Vance C, de Belleruche J, Baas F, Ten Asbroek ALMA, Muñoz-Blanco JL, Hernandez DG, Ding J, Gibbs JR, Scholz SW, Floeter MK, Campbell RH, Landi F, Bowser R, Pulst SM, Ravits JM, MacGowan DJL, Kirby J, Pioro EP, Pamphlett R, Broach J, Gerhard G, Dunckley TL, Brady CB, Kowall NW, Troncoso JC, Le Ber I, Mouzat K, Lumbroso S, Heiman-Patterson TD, Kamel F, Van Den Bosch L, Baloh RH, Strom TM, Meitinger T, Shatunov A, Van Eijk KR, de Carvalho M, Kooyman M, Middelkoop B, Moisse M, McLaughlin RL, Van Es MA, Weber M, Boylan KB, Van Blitterswijk M, Rademakers R, Morrison KE, Basak AN, Mora JS, Drory VE, Shaw PJ, Turner MR, Talbot K, Hardiman O, Williams KL, Fifita JA, Nicholson GA, Blair IP, Rouleau GA, Esteban-Pérez J, García-Redondo A, Al-Chalabi A; Project MinE ALS Sequencing Consortium, Rogaeva E, Zinman L, Ostrow LW, Maragakis NJ, Rothstein JD, Simmons Z, Cooper-Knock J, Brice A, Goutman SA, Feldman EL, Gibson SB, Taroni F, Ratti A, Gellera C, Van Damme P, Robberecht W, Fratta P, Sabatelli M, Lunetta C, Ludolph AC, Andersen PM, Weishaupt JH, Camu W, Trojanowski JQ, Van Deerlin VM, Brown RH Jr, van den Berg LH, Veldink JH, Harms MB, Glass JD, Stone DJ, Tienari P, Silani V, Chiò A, Shaw CE, Traynor BJ, Landers JE. Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*. 2018 Mar 21;97(6):1268-1283.e6. doi: 10.1016/j.neuron.2018.02.027. PMID: 29566793; PMCID: PMC5867896.
32. Perez Carrion M, Pischedda F, Biosa A, Russo I, Straniero L, Civiero L, Guida M, Gloeckner CJ, Ticozzi N, Tiloca C, Mariani C, Pezzoli G, Duga S, Pichler I, Pan L, Landers JE, Greggio E, Hess MW, Goldwurm S, Piccoli G. The LRRK2 Variant E193K Prevents Mitochondrial Fission Upon MPP+ Treatment by Altering LRRK2 Binding to DRP1. *Front Mol Neurosci*. 2018 Feb 28;11:64. doi: 10.3389/fnmol.2018.00064. PMID: 29541021; PMCID: PMC5835904.
 33. Mandrioli J, Ferri L, Fasano A, Zucchi E, Fini N, Moglia C, Lunetta C, Marinou K, Ticozzi N, Drago Ferrante G, Scialo C, Sorarù G, Trojsi F, Conte A, Falzone YM, Tortelli R, Russo M, Sansone VA, Mora G, Silani V, Volanti P, Caponnetto C, Querin G, Monsurrò MR, Sabatelli M, Chiò A, Riva N, Logroscino G, Messina S, Calvo A. Cardiovascular diseases may play a negative role in the prognosis of amyotrophic lateral sclerosis. *Eur J Neurol*. 2018 Jun;25(6):861-868. doi: 10.1111/ene.13620. Epub 2018 Apr 15. PMID: 29512869.
 34. Corrado L, Tiloca C, Locci C, Bagarotti A, Hamzeiy H, Colombrita C, De Marchi F, Barizzone N, Cotella D, Ticozzi N, Mazzini L, Nazli Basak A, Ratti A, Silani V, D'Alfonso S. Characterization of the c9orf72 GC-rich low complexity sequence in two cohorts of Italian and Turkish ALS cases. *Amyotroph Lateral Scler Frontotemporal Degener*. 2018 Aug;19(5-6):426-431. doi: 10.1080/21678421.2018.1440407. Epub 2018 Feb 28. PMID: 29490503.
 35. Hamzeiy H, Savaş D, Tunca C, Şen NE, Gündoğdu Eken A, Şahbaz I, Calini D, Tiloca C, Ticozzi N, Ratti A, Silani V, Başak AN. Elevated Global DNA Methylation Is Not Exclusive to Amyotrophic Lateral Sclerosis and Is Also Observed in Spinocerebellar Ataxia Types 1 and 2. *Neurodegener Dis*. 2018;18(1):38-48. doi: 10.1159/000486201. Epub 2018 Feb 9. PMID: 29428949.
 36. van der Spek RA, van Rheenen W, Pulit SL, Kenna KP, Ticozzi N, Kooyman M, McLaughlin RL, Moisse M, van Eijk KR, van Vugt JJFA, Iacoangeli A, Andersen P, Nazli Basak A, Blair I, de Carvalho M, Chio A, Corcia P, Couratier P, Drory VE, Glass JD, Hardiman O, Mora JS, Morrison KE, Mitne-Neto M, Robberecht W, Shaw PJ, Panadés MP, van Damme P, Silani V, Gotkine M, Weber M, van Es MA, Landers JE, Al-Chalabi A, van den Berg LH, Veldink JH; PROJECT MINE ALS SEQUENCING CONSORTIUM. Reconsidering the causality of TIA1 mutations in ALS. *Amyotroph Lateral Scler Frontotemporal Degener*. 2018 Feb;19(1-2):1-3. doi: 10.1080/21678421.2017.1413118. Epub 2017 Dec 13. Erratum in: *Amyotroph Lateral Scler Frontotemporal Degener*. 2018 Feb;19(1-2):161. PMID: 29235362; PMCID: PMC6516059.
 37. Cooper-Knock J, Robins H, Niedermoser I, Wyles M, Heath PR, Higginbottom A, Walsh T, Kazoka M; Project MinE ALS Sequencing Consortium, Ince PG, Hautbergue GM, McDermott CJ, Kirby J, Shaw PJ. Targeted Genetic Screen in Amyotrophic Lateral Sclerosis Reveals Novel Genetic Variants with Synergistic Effect on Clinical Phenotype. *Front Mol Neurosci*. 2017 Nov 9;10:370. doi: 10.3389/fnmol.2017.00370. PMID: 29170628; PMCID: PMC5684183.
 38. Trojsi F, Siciliano M, Femiano C, Santangelo G, Lunetta C, Calvo A, Moglia C, Marinou K, Ticozzi N, Drago Ferrante G, Scialò C, Sorarù G, Conte A, Falzone YM, Tortelli R, Russo M, Sansone VA, Chiò A, Mora G, Poletti B, Volanti P, Caponnetto C, Querin G, Sabatelli M, Riva N, Logroscino G, Messina S, Fasano A, Monsurrò MR, Tedeschi G, Mandrioli J. Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. *J Neurol*. 2017 Nov;264(11):2224-2231. doi: 10.1007/s00415-017-8619-4. Epub 2017 Sep 15. PMID: 28914354.
 39. Carelli L, Solca F, Faini A, Meriggi P, Sangalli D, Cipresso P, Riva G, Ticozzi N, Ciammola A, Silani V, Poletti B. Brain-Computer Interface for Clinical Purposes: Cognitive Assessment and Rehabilitation. *Biomed Res Int*. 2017;2017:1695290. doi: 10.1155/2017/1695290. Epub 2017 Aug 23. PMID: 28913349; PMCID: PMC5587953.
 40. Banfi P, Volpato E, Valota C, D'Ascenzo S, Alunno CB, Lax A, Nicolini A, Ticozzi N, Silani V, Bach JR. Use of Noninvasive Ventilation During Feeding Tube Placement. *Respir Care*. 2017 Nov;62(11):1474-1484. doi: 10.4187/respcare.05031. Epub 2017 Aug 14. PMID: 28807987.
 41. Bonati MT, Verde F, Hladnik U, Cattelan P, Campana L, Castronovo C, Ticozzi N, Maderna L, Colombrita C, Papa S, Banfi P, Silani V. A novel nonsense *ATP7A* pathogenic variant in a family exhibiting a variable occipital

- horn syndrome phenotype. *Mol Genet Metab Rep*. 2017 Jul 21;13:14-17. doi: 10.1016/j.ymgmr.2017.07.007. PMID: 28761814; PMCID: PMC5522958.
42. van Doormaal PTC, Ticozzi N, Weishaupt JH, Kenna K, Diekstra FP, Verde F, Andersen PM, Dekker AM, Tiloca C, Marroquin N, Overste DJ, Pensato V, Nürnberg P, Pulit SL, Schellevis RD, Calini D, Altmüller J, Francioli LC, Muller B, Castellotti B, Motameny S, Ratti A, Wolf J, Gellera C, Ludolph AC, van den Berg LH, Kubisch C, Landers JE, Veldink JH, Silani V, Volk AE. The role of de novo mutations in the development of amyotrophic lateral sclerosis. *Hum Mutat*. 2017 Nov;38(11):1534-1541. doi: 10.1002/humu.23295. Epub 2017 Aug 3. PMID: 28714244; PMCID: PMC6599399.
 43. Poletti B, Carelli L, Solca F, Lafronza A, Pedroli E, Faini A, Ticozzi N, Ciammola A, Meriggi P, Cipresso P, Lulé D, Ludolph AC, Riva G, Silani V. An eye-tracker controlled cognitive battery: overcoming verbal-motor limitations in ALS. *J Neurol*. 2017 Jun;264(6):1136-1145. doi: 10.1007/s00415-017-8506-z. Epub 2017 May 13. PMID: 28503706.
 44. Smith BN, Topp SD, Fallini C, Shibata H, Chen HJ, Troakes C, King A, Ticozzi N, Kenna KP, Soragia-Gkazi A, Miller JW, Sato A, Dias DM, Jeon M, Vance C, Wong CH, de Majo M, Kattuah W, Mitchell JC, Scotter EL, Parkin NW, Sapp PC, Nolan M, Nestor PJ, Simpson M, Weale M, Lek M, Baas F, Vianney de Jong JM, Ten Asbroek ALMA, Redondo AG, Esteban-Pérez J, Tiloca C, Verde F, Duga S, Leigh N, Pall H, Morrison KE, Al-Chalabi A, Shaw PJ, Kirby J, Turner MR, Talbot K, Hardiman O, Glass JD, De Belleruche J, Maki M, Moss SE, Miller C, Gellera C, Ratti A, Al-Sarraj S, Brown RH Jr, Silani V, Landers JE, Shaw CE. Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. *Sci Transl Med*. 2017 May 3;9(388):eaad9157. doi: 10.1126/scitranslmed.aad9157. PMID: 28469040; PMCID: PMC6599403.
 45. McLaughlin RL, Schijven D, van Rheenen W, van Eijk KR, O'Brien M, Kahn RS, Ophoff RA, Goris A, Bradley DG, Al-Chalabi A, van den Berg LH, Luykx JJ, Hardiman O, Veldink JH; Project MinE GWAS Consortium; Schizophrenia Working Group of the Psychiatric Genomics Consortium. Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. *Nat Commun*. 2017 Mar 21;8:14774. doi: 10.1038/ncomms14774. PMID: 28322246; PMCID: PMC5364411.
 46. Poletti B, Carelli L, Solca F, Lafronza A, Pedroli E, Faini A, Zago S, Ticozzi N, Ciammola A, Morelli C, Meriggi P, Cipresso P, Lulé D, Ludolph AC, Riva G, Silani V. An eye-tracking controlled neuropsychological battery for cognitive assessment in neurological diseases. *Neurol Sci*. 2017 Apr;38(4):595-603. doi: 10.1007/s10072-016-2807-3. Epub 2017 Jan 11. PMID: 28078566.
 47. Calvo A, Moglia C, Lunetta C, Marinou K, Ticozzi N, Ferrante GD, Scialo C, Sorarù G, Trojsi F, Conte A, Falzone YM, Tortelli R, Russo M, Chiò A, Sansone VA, Mora G, Silani V, Volanti P, Caponnetto C, Querin G, Monsurro MR, Sabatelli M, Riva N, Logroscino G, Messina S, Fini N, Mandrioli J. Factors predicting survival in ALS: a multicenter Italian study. *J Neurol*. 2017 Jan;264(1):54-63. doi: 10.1007/s00415-016-8313-y. Epub 2016 Oct 24. PMID: 27778156.
 48. van Rheenen W, Shatunov A, Dekker AM, McLaughlin RL, Diekstra FP, Pulit SL, van der Spek RA, Vösa U, de Jong S, Robinson MR, Yang J, Fogh I, van Doormaal PT, Tazelaar GH, Koppers M, Blokhuis AM, Sproviero W, Jones AR, Kenna KP, van Eijk KR, Harschnitz O, Schellevis RD, Brands WJ, Medic J, Menelaou A, Vajda A, Ticozzi N, Lin K, Rogelj B, Vrabec K, Ravnik-Glavač M, Koritnik B, Zidar J, Leonardis L, Grošelj LD, Millicamps S, Salachas F, Meininger V, de Carvalho M, Pinto S, Mora JS, Rojas-García R, Polak M, Chandran S, Colville S, Swingle R, Morrison KE, Shaw PJ, Hardy J, Orrell RW, Pittman A, Sidle K, Fratta P, Malaspina A, Topp S, Petri S, Abdulla S, Drepper C, Sendtner M, Meyer T, Ophoff RA, Staats KA, Wiedau-Pazos M, Lomen-Hoerth C, Van Deerlin VM, Trojanowski JQ, Elman L, McCluskey L, Basak AN, Tunca C, Hamzeiy H, Parman Y, Meitinger T, Lichtner P, Radivojkov-Blagojevic M, Andres CR, Maurel C, Bensimon G, Landwehrmeyer B, Brice A, Payan CA, Saker-Delye S, Dürr A, Wood NW, Tittmann L, Lieb W, Franke A, Rietschel M, Cichon S, Nöthen MM, Amouyel P, Tzourio C, Dartigues JF, Uitterlinden AG, Rivadeneira F, Estrada K, Hofman A, Curtis C, Blauw HM, van der Kooij AJ, de Visser M, Goris A, Weber M, Shaw CE, Smith BN, Pansarasa O, Cereda C, Del Bo R, Comi GP, D'Alfonso S, Bertolin C, Sorarù G, Mazzini L, Pensato V, Gellera C, Tiloca C, Ratti A, Calvo A, Moglia C, Brunetti M, Arcuti S, Capozzo R, Zecca C, Lunetta C, Penco S, Riva N, Padovani A, Filosto M, Muller B, Stuit RJ; PARALS Registry; SLALOM Group; SLAP Registry; FALS Sequencing Consortium; SLAGEN Consortium; NNIPPS Study Group, Blair I, Zhang K, McCann EP, Fifita JA, Nicholson GA, Rowe DB, Pamphlett R, Kiernan MC, Grosskreutz J, Witte OW, Ringer T, Prell T, Stubendorff B, Kurth I, Hübner CA, Leigh PN, Casale F, Chio A, Beghi E, Pupillo E, Tortelli R, Logroscino G, Powell J, Ludolph AC, Weishaupt JH, Robberecht W, Van Damme P, Franke L, Pers TH, Brown RH, Glass JD, Landers JE, Hardiman O, Andersen PM, Corcia P, Vourc'h P, Silani V, Wray NR, Visscher PM, de Bakker PI, van Es MA, Pasterkamp RJ, Lewis CM, Breen G, Al-Chalabi A, van den Berg LH, Veldink JH. Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nat Genet*. 2016 Sep;48(9):1043-8. doi: 10.1038/ng.3622. Epub 2016 Jul 25. PMID: 27455348; PMCID: PMC5556360.
 49. Kenna KP, van Doormaal PT, Dekker AM, Ticozzi N, Kenna BJ, Diekstra FP, van Rheenen W, van Eijk KR, Jones AR, Keagle P, Shatunov A, Sproviero W, Smith BN, van Es MA, Topp SD, Kenna A, Miller JW, Fallini C, Tiloca C, McLaughlin RL, Vance C, Troakes C, Colombrita C, Mora G, Calvo A, Verde F, Al-Sarraj S, King A, Calini D, de Belleruche J, Baas F, van der Kooij AJ, de Visser M, Ten Asbroek AL, Sapp PC, McKenna-Yasek D, Polak M, Asress S, Muñoz-Blanco JL, Strom TM, Meitinger T, Morrison KE; SLAGEN Consortium, Lauria G, Williams KL, Leigh PN, Nicholson GA, Blair IP, Leblond CS, Dion PA, Rouleau GA, Pall H, Shaw PJ, Turner MR, Talbot K, Taroni F, Boylan KB, Van Blitterswijk M, Rademakers R, Esteban-Pérez J, Garcia-Redondo A, Van Damme P, Robberecht W, Chio A, Gellera C, Drepper C, Sendtner M, Ratti A, Glass JD, Mora JS, Basak NA, Hardiman O,

- Ludolph AC, Andersen PM, Weishaupt JH, Brown RH Jr, Al-Chalabi A, Silani V, Shaw CE, van den Berg LH, Veldink JH, Landers JE. NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nat Genet.* 2016 Sep;48(9):1037-42. doi: 10.1038/ng.3626. Epub 2016 Jul 25. PMID: 27455347; PMCID: PMC5560030.
50. Fogh I, Lin K, Tiloca C, Rooney J, Gellera C, Diekstra FP, Ratti A, Shatunov A, van Es MA, Proitsi P, Jones A, Sproviero W, Chiò A, McLaughlin RL, Sorarù G, Corrado L, Stahl D, Del Bo R, Cereda C, Castellotti B, Glass JD, Newhouse S, Dobson R, Smith BN, Topp S, van Rheenen W, Meininger V, Melki J, Morrison KE, Shaw PJ, Leigh PN, Andersen PM, Comi GP, Ticozzi N, Mazzini L, D'Alfonso S, Traynor BJ, Van Damme P, Robberecht W, Brown RH, Landers JE, Hardiman O, Lewis CM, van den Berg LH, Shaw CE, Veldink JH, Silani V, Al-Chalabi A, Powell J. Association of a Locus in the CAMTA1 Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. *JAMA Neurol.* 2016 Jul 1;73(7):812-20. doi: 10.1001/jamaneurol.2016.1114. PMID: 27244217; PMCID: PMC5556366.
 51. Poletti B, Solca F, Carelli L, Madotto F, Lafronza A, Faini A, Monti A, Zago S, Calini D, Tiloca C, Doretti A, Verde F, Ratti A, Ticozzi N, Abrahams S, Silani V. The validation of the Italian Edinburgh Cognitive and Behavioural ALS Screen (ECAS). *Amyotroph Lateral Scler Frontotemporal Degener.* 2016 Oct- Nov;17(7-8):489-498. doi: 10.1080/21678421.2016.1183679. Epub 2016 May 24. PMID: 27219526.
 52. Poletti B, Carelli L, Solca F, Lafronza A, Pedrolì E, Faini A, Zago S, Ticozzi N, Meriggi P, Cipresso P, Lulé D, Ludolph AC, Riva G, Silani V. Cognitive assessment in Amyotrophic Lateral Sclerosis by means of P300-Brain Computer Interface: a preliminary study. *Amyotroph Lateral Scler Frontotemporal Degener.* 2016 Oct-Nov;17(7-8):473-481. doi: 10.1080/21678421.2016.1181182. Epub 2016 May 12. PMID: 27169693.
 53. Williams KL, Topp S, Yang S, Smith B, Fifita JA, Warraich ST, Zhang KY, Farrarwell N, Vance C, Hu X, Chesi A, Leblond CS, Lee A, Rayner SL, Sundaramoorthy V, Dobson-Stone C, Molloy MP, van Blitterswijk M, Dickson DW, Petersen RC, Graff-Radford NR, Boeve BF, Murray ME, Pottier C, Don E, Winnick C, McCann EP, Hogan A, Daoud H, Levert A, Dion PA, Mitsui J, Ishiura H, Takahashi Y, Goto J, Kost J, Gellera C, Gkazi AS, Miller J, Stockton J, Brooks WS, Boundy K, Polak M, Muñoz-Blanco JL, Esteban-Pérez J, Rábano A, Hardiman O, Morrison KE, Ticozzi N, Silani V, de Bellerocche J, Glass JD, Kwok JB, Guillemin GJ, Chung RS, Tsuji S, Brown RH Jr, Garcia-Redondo A, Rademakers R, Landers JE, Gitler AD, Rouleau GA, Cole NJ, Yerbury JJ, Atkin JD, Shaw CE, Nicholson GA, Blair IP. CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nat Commun.* 2016 Apr 15;7:11253. doi: 10.1038/ncomms11253. PMID: 27080313; PMCID: PMC4835537.
 54. Verde F, Ticozzi N, Messina S, Calcagno N, Girotti F, Maderna L, Moda F, Scola E, Falini A, Tagliavini F, Silani V. MRI abnormalities found 1 year prior to symptom onset in a case of Creutzfeldt-Jakob disease. *J Neurol.* 2016 Mar;263(3):597-9. doi: 10.1007/s00415-016-8022-6. Epub 2016 Feb 12. PMID: 26872662.
 55. Benedetti MD, Pugliatti M, D'Alessandro R, Beghi E, Chiò A, Logroscino G, Filippini G, Galeotti F, Massari M, Santuccio C, Raschetti R; ITANG Study Group. A Multicentric Prospective Incidence Study of Guillain-Barré Syndrome in Italy. *The ITANG Study. Neuroepidemiology.* 2015;45(2):90-9. doi: 10.1159/000438752. Epub 2015 Aug 29. PMID: 26329724.
 56. Pensato V, Tiloca C, Corrado L, Bertolin C, Sardone V, Del Bo R, Calini D, Mandrioli J, Lauria G, Mazzini L, Querin G, Ceroni M, Cantello R, Corti S, Castellotti B, Soldà G, Duga S, Comi GP, Cereda C, Sorarù G, D'Alfonso S, Taroni F, Shaw CE, Landers JE, Ticozzi N, Ratti A, Gellera C, Silani V; SLAGEN Consortium. TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. *J Neurol.* 2015 May;262(5):1376-8. doi: 10.1007/s00415-015-7739-y. Epub 2015 Apr 18. PMID: 25893256; PMCID: PMC6614739.
 57. Cirulli ET, Lasseigne BN, Petrovski S, Sapp PC, Dion PA, Leblond CS, Couthouis J, Lu YF, Wang Q, Krueger BJ, Ren Z, Keebler J, Han Y, Levy SE, Boone BE, Wimbish JR, Waite LL, Jones AL, Carulli JP, Day-Williams AG, Staropoli JF, Xin WW, Chesi A, Raphael AR, McKenna-Yasek D, Cady J, Vianney de Jong JM, Kenna KP, Smith BN, Topp S, Miller J, Gkazi A; FALS Sequencing Consortium, Al-Chalabi A, van den Berg LH, Veldink J, Silani V, Ticozzi N, Shaw CE, Baloh RH, Appel S, Simpson E, Lagier-Tourenne C, Pulst SM, Gibson S, Trojanowski JQ, Elman L, McCluskey L, Grossman M, Shneider NA, Chung WK, Ravits JM, Glass JD, Sims KB, Van Deerlin VM, Maniatis T, Hayes SD, Ordureau A, Swarup S, Landers J, Baas F, Allen AS, Bedlack RS, Harper JW, Gitler AD, Rouleau GA, Brown R, Harms MB, Cooper GM, Harris T, Myers RM, Goldstein DB. Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. *Science.* 2015 Mar 27;347(6229):1436-41. doi: 10.1126/science.aaa3650. Epub 2015 Feb 19. PMID: 25700176; PMCID: PMC4437632.
 58. Ronchi D, Riboldi G, Del Bo R, Ticozzi N, Scarlato M, Galimberti D, Corti S, Silani V, Bresolin N, Comi GP. CHCHD10 mutations in Italian patients with sporadic amyotrophic lateral sclerosis. *Brain.* 2015 Aug;138(Pt 8):e372. doi: 10.1093/brain/awu384. Epub 2015 Jan 8. PMID: 25576308.
 59. Smith BN, Vance C, Scotter EL, Troakes C, Wong CH, Topp S, Maekawa S, King A, Mitchell JC, Lund K, Al-Chalabi A, Ticozzi N, Silani V, Sapp P, Brown RH Jr, Landers JE, Al-Sarraj S, Shaw CE. Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. *Neurobiol Aging.* 2015 Mar;36(3):1602.e17-27. doi: 10.1016/j.neurobiolaging.2014.10.032. Epub 2014 Oct 31. PMID: 25499087; PMCID: PMC4357530.
 60. Smith BN, Ticozzi N, Fallini C, Gkazi AS, Topp S, Kenna KP, Scotter EL, Kost J, Keagle P, Miller JW, Calini D, Vance C, Danielson EW, Troakes C, Tiloca C, Al-Sarraj S, Lewis EA, King A, Colombrina C, Pensato V, Castellotti B, de Bellerocche J, Baas F, ten Asbroek AL, Sapp PC, McKenna-Yasek D, McLaughlin RL, Polak M, Asress S, Esteban-Pérez J, Muñoz-Blanco JL, Simpson M; SLAGEN Consortium, van Rheenen W, Diekstra FP, Lauria G, Duga S, Corti S, Cereda C, Corrado L, Sorarù G, Morrison KE, Williams KL, Nicholson GA, Blair IP, Dion PA, Leblond CS, Rouleau GA, Hardiman O, Veldink JH, van den Berg LH, Al-Chalabi A, Pall H, Shaw PJ, Turner MR,

- Talbot K, Taroni F, García-Redondo A, Wu Z, Glass JD, Gellera C, Ratti A, Brown RH Jr, Silani V, Shaw CE, Landers JE. Exome-wide rare variant analysis identifies TUBA4A mutations associated with familial ALS. *Neuron*. 2014 Oct 22;84(2):324-31. doi: 10.1016/j.neuron.2014.09.027. Epub 2014 Oct 22. PMID: 25374358; PMCID: PMC4521390.
61. Banfi P, Ticozzi N, Lax A, Guidugli GA, Nicolini A, Silani V. A review of options for treating sialorrhea in amyotrophic lateral sclerosis. *Respir Care*. 2015 Mar;60(3):446-54. doi: 10.4187/respcare.02856. Epub 2014 Sep 16. PMID: 25228780.
 62. van Doormaal PT, Ticozzi N, Gellera C, Ratti A, Taroni F, Chiò A, Calvo A, Mora G, Restagno G, Traynor BJ, Birve A, Lemmens R, van Es MA, Saris CG, Blauw HM, van Vught PW, Groen EJ, Corrado L, Mazzini L, Del Bo R, Corti S, Waibel S, Meyer T, Ludolph AC, Goris A, van Damme P, Robberecht W, Shatunov A, Fogh I, Andersen PM, D'Alfonso S, Hardiman O, Cronin S, Rujescu D, Al-Chalabi A, Landers JE, Silani V, van den Berg LH, Veldink JH. Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. *Neurobiol Aging*. 2014 Oct;35(10):2420.e13-4. doi: 10.1016/j.neurobiolaging.2014.04.014. Epub 2014 Apr 19. PMID: 24838185; PMCID: PMC5496711.
 63. Fogh I, Ratti A, Gellera C, Lin K, Tiloca C, Moskvina V, Corrado L, Sorarù G, Cereda C, Corti S, Gentilini D, Calini D, Castellotti B, Mazzini L, Querin G, Gagliardi S, Del Bo R, Conforti FL, Siciliano G, Inghilleri M, Saccà F, Bongioanni P, Penco S, Corbo M, Sorbi S, Filosto M, Ferlini A, Di Blasio AM, Signorini S, Shatunov A, Jones A, Shaw PJ, Morrison KE, Farmer AE, Van Damme P, Robberecht W, Chiò A, Traynor BJ, Sendtner M, Melki J, Meininger V, Hardiman O, Andersen PM, Leigh NP, Glass JD, Overste D, Diekstra FP, Veldink JH, van Es MA, Shaw CE, Weale ME, Lewis CM, Williams J, Brown RH, Landers JE, Ticozzi N, Ceroni M, Pegoraro E, Comi GP, D'Alfonso S, van den Berg LH, Taroni F, Al-Chalabi A, Powell J, Silani V; SLAGEN Consortium and Collaborators. A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. *Hum Mol Genet*. 2014 Apr 15;23(8):2220-31. doi: 10.1093/hmg/ddt587. Epub 2013 Nov 20. PMID: 24256812; PMCID: PMC3959809.
 64. Ticozzi N, Tiloca C, Calini D, Gagliardi S, Altieri A, Colombrita C, Cereda C, Ratti A, Pezzoli G, Borroni B, Goldwurm S, Padovani A, Silani V. C9orf72 repeat expansions are restricted to the ALS-FTD spectrum. *Neurobiol Aging*. 2014 Apr;35(4):936.e13-7. doi: 10.1016/j.neurobiolaging.2013.09.037. Epub 2013 Oct 2. PMID: 24169076.
 65. Calini D, Corrado L, Del Bo R, Gagliardi S, Pensato V, Verde F, Corti S, Mazzini L, Milani P, Castellotti B, Bertolin C, Sorarù G, Cereda C, Comi GP, D'Alfonso S, Gellera C, Ticozzi N, Landers JE, Ratti A, Silani V; SLAGEN Consortium. Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. *Neurobiol Aging*. 2013 Nov;34(11):2695.e11-2. doi: 10.1016/j.neurobiolaging.2013.05.025. Epub 2013 Jul 2. PMID: 23827524; PMCID: PMC6591723.
 66. Galeotti F, Massari M, D'Alessandro R, Beghi E, Chiò A, Logroscino G, Filippini G, Benedetti MD, Pugliatti M, Santuccio C, Raschetti R; ITANG study group. Risk of Guillain-Barré syndrome after 2010-2011 influenza vaccination. *Eur J Epidemiol*. 2013 May;28(5):433-44. doi: 10.1007/s10654-013-9797-8. Epub 2013 Mar 31. PMID: 23543123; PMCID: PMC3672511.
 67. Beghi E, Pupillo E, Bonito V, Buzzi P, Caponnetto C, Chiò A, Corbo M, Giannini F, Inghilleri M, Bella VL, Logroscino G, Lorusso L, Lunetta C, Mazzini L, Messina P, Mora G, Perini M, Quadrelli ML, Silani V, Simone IL, Tremolizzo L; Italian ALS Study Group. Randomized double-blind placebo-controlled trial of acetyl-L-carnitine for ALS. *Amyotroph Lateral Scler Frontotemporal Degener*. 2013 Sep;14(5-6):397-405. doi: 10.3109/21678421.2013.764568. Epub 2013 Feb 19. PMID: 23421600.
 68. Gellera C, Tiloca C, Del Bo R, Corrado L, Pensato V, Agostini J, Cereda C, Ratti A, Castellotti B, Corti S, Bagarotti A, Cagnin A, Milani P, Gabelli C, Riboldi G, Mazzini L, Sorarù G, D'Alfonso S, Taroni F, Comi GP, Ticozzi N, Silani V; SLAGEN Consortium. Ubiquilin 2 mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. *J Neurol Neurosurg Psychiatry*. 2013 Feb;84(2):183-7. doi: 10.1136/jnnp-2012-303433. Epub 2012 Nov 8. PMID: 23138764.
 69. Tiloca C, Ticozzi N, Pensato V, Corrado L, Del Bo R, Bertolin C, Fenoglio C, Gagliardi S, Calini D, Lauria G, Castellotti B, Bagarotti A, Corti S, Galimberti D, Cagnin A, Gabelli C, Ranieri M, Ceroni M, Siciliano G, Mazzini L, Cereda C, Scarpini E, Sorarù G, Comi GP, D'Alfonso S, Gellera C, Ratti A, Landers JE, Silani V; SLAGEN Consortium. Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. *Neurobiol Aging*. 2013 May;34(5):1517.e9-10. doi: 10.1016/j.neurobiolaging.2012.09.016. Epub 2012 Oct 11. PMID: 23063648; PMCID: PMC3548975.
 70. Wu CH, Fallini C, Ticozzi N, Keagle PJ, Sapp PC, Piotrowska K, Lowe P, Koppers M, McKenna-Yasek D, Baron DM, Kost JE, Gonzalez-Perez P, Fox AD, Adams J, Taroni F, Tiloca C, Leclerc AL, Chafe SC, Mangroo D, Moore MJ, Zitzewitz JA, Xu ZS, van den Berg LH, Glass JD, Siciliano G, Cirulli ET, Goldstein DB, Salachas F, Meininger V, Rossoll W, Ratti A, Gellera C, Bosco DA, Bassell GJ, Silani V, Drory VE, Brown RH Jr, Landers JE. Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. *Nature*. 2012 Aug 23;488(7412):499-503. doi: 10.1038/nature11280. PMID: 22801503; PMCID: PMC3575525.
 71. Ratti A, Corrado L, Castellotti B, Del Bo R, Fogh I, Cereda C, Tiloca C, D'Ascenzo C, Bagarotti A, Pensato V, Ranieri M, Gagliardi S, Calini D, Mazzini L, Taroni F, Corti S, Ceroni M, Oggioni GD, Lin K, Powell JF, Sorarù G, Ticozzi N, Comi GP, D'Alfonso S, Gellera C, Silani V; SLAGEN Consortium. C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. *Neurobiol Aging*. 2012 Oct;33(10):2528.e7-14. doi: 10.1016/j.neurobiolaging.2012.06.008. Epub 2012 Jul 4. PMID: 22766072.

72. Ticozzi N, Tiloca C, Mencacci NE, Morelli C, Doretti A, Rusconi D, Colombrita C, Sangalli D, Verde F, Finelli P, Messina S, Ratti A, Silani V. Oligoclonal bands in the cerebrospinal fluid of amyotrophic lateral sclerosis patients with disease-associated mutations. *J Neurol.* 2013 Jan;260(1):85-92. doi: 10.1007/s00415-012-6589-0. Epub 2012 Jul 1. PMID: 22752089; PMCID: PMC4196642.
73. Smith BN, Newhouse S, Shatunov A, Vance C, Topp S, Johnson L, Miller J, Lee Y, Troakes C, Scott KM, Jones A, Gray I, Wright J, Hortobágyi T, Al-Sarraj S, Rogelj B, Powell J, Lupton M, Lovestone S, Sapp PC, Weber M, Nestor PJ, Schelhaas HJ, Asbroek AA, Silani V, Gellera C, Taroni F, Ticozzi N, Van den Berg L, Veldink J, Van Damme P, Robberecht W, Shaw PJ, Kirby J, Pall H, Morrison KE, Morris A, de Bellerocche J, Vianney de Jong JM, Baas F, Andersen PM, Landers J, Brown RH Jr, Weale ME, Al-Chalabi A, Shaw CE. The C9ORF72 expansion mutation is a common cause of ALS+/-FTD in Europe and has a single founder. *Eur J Hum Genet.* 2013 Jan;21(1):102-8. doi: 10.1038/ejhg.2012.98. Epub 2012 Jun 13. PMID: 22692064; PMCID: PMC3522204.
74. Gellera C, Ticozzi N, Pensato V, Nanetti L, Castucci A, Castellotti B, Lauria G, Taroni F, Silani V, Mariotti C. ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. *Neurobiol Aging.* 2012 Aug;33(8):1847.e15-21. doi: 10.1016/j.neurobiolaging.2012.02.004. Epub 2012 Mar 16. PMID: 22425256.
75. Ramos EM, Keagle P, Gillis T, Lowe P, Mysore JS, Leclerc AL, Ratti A, Ticozzi N, Gellera C, Gusella JF, Silani V, Alonso I, Brown RH Jr, MacDonald ME, Landers JE. Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. *Amyotroph Lateral Scler.* 2012 May;13(3):265-9. doi: 10.3109/17482968.2011.653573. Epub 2012 Mar 13. PMID: 22409360.
76. van Es MA, Schelhaas HJ, van Vught PW, Ticozzi N, Andersen PM, Groen EJ, Schulte C, Blauw HM, Koppers M, Diekstra FP, Fumoto K, LeClerc AL, Keagle P, Bloem BR, Scheffer H, van Nuenen BF, van Blitterswijk M, van Rheenen W, Wills AM, Lowe PP, Hu GF, Yu W, Kishikawa H, Wu D, Folkerth RD, Mariani C, Goldwurm S, Pezzoli G, Van Damme P, Lemmens R, Dahlberg C, Birve A, Fernández-Santiago R, Waibel S, Klein C, Weber M, van der Kooij AJ, de Visser M, Verbaan D, van Hilten JJ, Heutink P, Hennekam EA, Cuppen E, Berg D, Brown RH Jr, Silani V, Gasser T, Ludolph AC, Robberecht W, Ophoff RA, Veldink JH, Pasterkamp RJ, de Bakker PI, Landers JE, van de Warrenburg BP, van den Berg LH. Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. *Ann Neurol.* 2011 Dec;70(6):964-73. doi: 10.1002/ana.22611. PMID: 22190368; PMCID: PMC5560057.
77. Tiloca C, Ratti A, Pensato V, Castucci A, Sorarù G, Del Bo R, Corrado L, Cereda C, D'Ascenzo C, Comi GP, Mazzini L, Castellotti B, Ticozzi N, Gellera C, Silani V; SLAGEN Consortium. Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. *Neurobiol Aging.* 2012 Mar;33(3):630.e1-2. doi: 10.1016/j.neurobiolaging.2011.10.025. Epub 2011 Dec 3. PMID: 22137929.
78. Del Bo R, Tiloca C, Pensato V, Corrado L, Ratti A, Ticozzi N, Corti S, Castellotti B, Mazzini L, Sorarù G, Cereda C, D'Alfonso S, Gellera C, Comi GP, Silani V; SLAGEN Consortium. Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. *J Neurol Neurosurg Psychiatry.* 2011 Nov;82(11):1239-43. doi: 10.1136/jnnp.2011.242313. Epub 2011 May 25. PMID: 21613650.
79. Savoiardo M, Erbetta A, Di Francesco JC, Brioschi M, Silani V, Falini A, Storchi G, Brighina L, Ferrarese C, Ticozzi N, Messina S, Girotti F. Cerebral amyloid angiopathy-related inflammation: an emerging disease. *Neuroradiol J.* 2011 May 15;24(2):253-7. doi: 10.1177/197140091102400214. Epub 2011 May 11. PMID: 24059616.
80. Ticozzi N, Vance C, Leclerc AL, Keagle P, Glass JD, McKenna-Yasek D, Sapp PC, Silani V, Bosco DA, Shaw CE, Brown RH Jr, Landers JE. Mutational analysis reveals the FUS homolog TAF15 as a candidate gene for familial amyotrophic lateral sclerosis. *Am J Med Genet B Neuropsychiatr Genet.* 2011 Apr;156B(3):285-90. doi: 10.1002/ajmg.b.31158. Epub 2011 Jan 13. PMID: 21438137.
81. Colombrita C, Onesto E, Tiloca C, Ticozzi N, Silani V, Ratti A. RNA-binding proteins and RNA metabolism: a new scenario in the pathogenesis of Amyotrophic lateral sclerosis. *Arch Ital Biol.* 2011 Mar;149(1):83-99. doi: 10.4449/aib.v149i1.1261. PMID: 21412718.
82. Ticozzi N, Tiloca C, Morelli C, Colombrita C, Poletti B, Doretti A, Maderna L, Messina S, Ratti A, Silani V. Genetics of familial Amyotrophic lateral sclerosis. *Arch Ital Biol.* 2011 Mar;149(1):65-82. doi: 10.4449/aib.v149i1.1262. PMID: 21412717.
83. Silani V, Messina S, Poletti B, Morelli C, Doretti A, Ticozzi N, Maderna L. The diagnosis of Amyotrophic lateral sclerosis in 2010. *Arch Ital Biol.* 2011 Mar;149(1):5-27. doi: 10.4449/aib.v149i1.1260. PMID: 21412713.
84. Corrado L, Gagliardi S, Carlomagno Y, Mennini T, Ticozzi N, Mazzini L, Silani V, Cereda C, D'Alfonso S. VPS54 genetic analysis in ALS Italian cohort. *Eur J Neurol.* 2011 Apr;18(4):e41-2. doi: 10.1111/j.1468-1331.2010.03260.x. Epub 2010 Nov 19. PMID: 21087364.
85. Ticozzi N, LeClerc AL, Keagle PJ, Glass JD, Wills AM, van Blitterswijk M, Bosco DA, Rodriguez-Leyva I, Gellera C, Ratti A, Taroni F, McKenna-Yasek D, Sapp PC, Silani V, Furlong CE, Brown RH Jr, Landers JE. Paraoxonase gene mutations in amyotrophic lateral sclerosis. *Ann Neurol.* 2010 Jul;68(1):102-7. doi: 10.1002/ana.21993. PMID: 20582942; PMCID: PMC2945725.
86. Ticozzi N, Ratti A, Silani V. Protein aggregation and defective RNA metabolism as mechanisms for motor neuron damage. *CNS Neurol Disord Drug Targets.* 2010 Jul;9(3):285-96. doi: 10.2174/187152710791292585. PMID: 20406182.
87. Silani V, Calzarossa C, Cova L, Ticozzi N. Stem cells in amyotrophic lateral sclerosis: motor neuron protection or replacement? *CNS Neurol Disord Drug Targets.* 2010 Jul;9(3):314-24. doi: 10.2174/187152710791292666. PMID: 20406179.

88. Ticozzi N, LeClerc AL, van Blitterswijk M, Keagle P, McKenna-Yasek DM, Sapp PC, Silani V, Wills AM, Brown RH Jr, Landers JE. Mutational analysis of TARDBP in neurodegenerative diseases. *Neurobiol Aging*. 2011 Nov;32(11):2096-9. doi: 10.1016/j.neurobiolaging.2009.11.018. Epub 2009 Dec 23. PMID: 20031275; PMCID: PMC2889148.
89. Ticozzi N, Silani V, LeClerc AL, Keagle P, Gellera C, Ratti A, Taroni F, Kwiatkowski TJ Jr, McKenna-Yasek DM, Sapp PC, Brown RH Jr, Landers JE. Analysis of FUS gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. *Neurology*. 2009 Oct 13;73(15):1180-5. doi: 10.1212/WNL.0b013e3181bbff05. Epub 2009 Sep 9. PMID: 19741215; PMCID: PMC2764725.
90. Fogh I, D'Alfonso S, Gellera C, Ratti A, Cereda C, Penco S, Corrado L, Sorarù G, Castellotti B, Tiloca C, Gagliardi S, Cozzi L, Lupton MK, Ticozzi N, Mazzini L, Shaw CE, Al-Chalabi A, Powell J, Silani V. No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. *Neurobiol Aging*. 2011 May;32(5):966-7. doi: 10.1016/j.neurobiolaging.2009.05.014. Epub 2009 Jun 13. PMID: 19525032.
91. Landers JE, Melki J, Meininger V, Glass JD, van den Berg LH, van Es MA, Sapp PC, van Vught PW, McKenna-Yasek DM, Blauw HM, Cho TJ, Polak M, Shi L, Wills AM, Broom WJ, Ticozzi N, Silani V, Ozoguz A, Rodriguez-Leyva I, Veldink JH, Ivinson AJ, Saris CG, Hosler BA, Barnes-Nessa A, Couture N, Wokke JH, Kwiatkowski TJ Jr, Ophoff RA, Cronin S, Hardiman O, Diekstra FP, Leigh PN, Shaw CE, Simpson CL, Hansen VK, Powell JF, Corcia P, Salachas F, Heath S, Galan P, Georges F, Horvitz HR, Lathrop M, Purcell S, Al-Chalabi A, Brown RH Jr. Reduced expression of the Kinesin-Associated Protein 3 (KIFAP3) gene increases survival in sporadic amyotrophic lateral sclerosis. *Proc Natl Acad Sci U S A*. 2009 Jun 2;106(22):9004-9. doi: 10.1073/pnas.0812937106. Epub 2009 May 18. PMID: 19451621; PMCID: PMC2683883.
92. Kwiatkowski TJ Jr, Bosco DA, Leclerc AL, Tamrazian E, Vanderburg CR, Russ C, Davis A, Gilchrist J, Kasarskis EJ, Munsat T, Valdmanis P, Rouleau GA, Hosler BA, Cortelli P, de Jong PJ, Yoshinaga Y, Haines JL, Pericak-Vance MA, Yan J, Ticozzi N, Siddique T, McKenna-Yasek D, Sapp PC, Horvitz HR, Landers JE, Brown RH Jr. Mutations in the FUS/TLS gene on chromosome 16 cause familial amyotrophic lateral sclerosis. *Science*. 2009 Feb 27;323(5918):1205-8. doi: 10.1126/science.1166066. PMID: 19251627.
93. Corrado L, Ratti A, Gellera C, Buratti E, Castellotti B, Carlomagno Y, Ticozzi N, Mazzini L, Testa L, Taroni F, Baralle FE, Silani V, D'Alfonso S. High frequency of TARDBP gene mutations in Italian patients with amyotrophic lateral sclerosis. *Hum Mutat*. 2009 Apr;30(4):688-94. doi: 10.1002/humu.20950. PMID: 19224587.
94. Gellera C, Colombrita C, Ticozzi N, Castellotti B, Bragato C, Ratti A, Taroni F, Silani V. Identification of new ANG gene mutations in a large cohort of Italian patients with amyotrophic lateral sclerosis. *Neurogenetics*. 2008 Feb;9(1):33-40. doi: 10.1007/s10048-007-0111-3. Epub 2007 Dec 18. PMID: 18087731.

MILANO, 14 SETTEMBRE 2021

NICOLA TICOZZI

Il sottoscritto è a conoscenza che, ai sensi dell'art. 76 del DPR 445/2000, le dichiarazioni mendaci, la falsità negli atti e l'uso di atti falsi sono puniti ai sensi del Codice penale e delle leggi speciali. Inoltre, il sottoscritto autorizza al trattamento dei dati personali ai sensi dell'art. 13 D. Lgs. 30 giugno 2003 n°196 – “Codice in materia di protezione dei dati personali” e dell'art. 13 GDPR 679/16 – “Regolamento europeo sulla protezione dei dati personali”