SANDRA D'ALFONSO

CV

Education

-1988: Bs.C degree in Biological Sciences 110/110, University of Turin, Italy.
-1995: Philosophical Doctorate in Human Genetics, University of Turin, Italy.
-1989-90 Short training visit at the National Cancer Institute, Amsterdam, the Netherlands
Current / past positions
1988-1991 Fellowship at Dept. Genetics Biology and Biochemistry, Turin, Italy
1996-2004 Researcher in Medical Genetics, Medical School of Novara, UPO (Università del Piemonte Orientale) University A. Avogadro, Novara
2004-2016 Associate Professor in Medical Genetics, Medical School of Novara, UPO (Università del Piemonte Orientale) University A. Avogadro, Novara
2016-now Full Professor in Medical Genetics, Medical School of Novara, UPO (Università del Piemonte Orientale) University A. Avogadro, Novara

Research Performance
ORCID: http://orcid.org/0000-0002-3983-9925
Publications: 210 papers in I.F. international journals
Metrics (Scopus# 55765749900): citations 12142, H-index: 59; H-index 2017-2022: 13.
Several presentations at National and International meetings and as invited speaker
Abstracts to congresses: over 280.
Research activities
Main field: genetics of neurodegenerative diseases and of autoimmune diseases
Specific domains:
Identification of genes associated to complex diseases (Multiple Sclerosis, Amyotrophic Lateral Sclerosis , Systemic Lupus Erythematosus, Celiac Disease) with a genomewide approach.
Identification of genes for familial forms neurodegenetative diseases (mainliy Amyotrophic Lateral Sclerosis) with next generation sequencing (WES, WGS). Genotype-phenotype correlation in neurodegenerative diseases. Association of genetic and genomic markers and environmental factors with multiple sclerosis clinical phenotype.
International Partnerships: Member of the IMSGC (International Multiple Sclerosis Genetics Consortium) Strategy Group, MultipleMS consortium, ALSgen consortium, SLAgen consortium, SLEgen consortium

Main achievements

The early research activity, starting in 1990, was initially focused on the immunogenetics of the HLA region and its association with autoimmune diseases and is documented by papers on international journals with high citations (D'Alfonso et al Immunogenetics, 1994,cit 365). Next she contributed as a principal investigator or co-authors to key papers identifying genes associated to autoimmune diseases (lupus:D'Alfonso et al.Arthritis Rheum. 2000; cit 100, 2005 cit 90, Alarcon et al. Nat Genet. 2008; cit

354; Multiple Sclerosis: IMSGC Nature 2011;, cit 1763, NatGen 2013; Cit 762, Nat Gen 2015 cit 141 Science 2019 cit 109).
Since 2006, she started a national and international collaboration on the genetics of ALS (Amyotrophic Lateral Sclerosis) and contributed as a principal investigator (Hum Mutat. 2009; cit 161, J Med Genet. 2010, cit 127) or co-authors to key studies to define the mutational spectrum of ALS patients in Italy and to identify new ALS causing genes (Science 2015, cit 509, Neuron 2018, cit 151), including studies focused on the role of tandem repeats, (Neurobiol Aging. 2015 Dec 8 , Neurology. 2015; Hum Genet. 2011)

At present, the major aims of the research activities, supported by national and international grants, are focused to 1) unravel the missing heritabiity of ALS and other neurodegenerative and neurological diseases by exploring the coding and non coding genomic regions analysing also complex sequence variations and integrating genomic with other omics data 2) analyse the effect of interaction of genomic sequence variations with life style and environmental factors in the susceptibility to neurological diseases (multiple sclerosis and ALS) and their clinical phenotypes 3) dissect the pathogenetic mechanisms underlying the association signals deriving from large genome wide association studies (GWAS) in multiple sclerosis

Funding (last ten years)

I have secured an independent grant portfolio of >2.7 M€ with the following grants:
- Unveiling the hidden side of NEUrodevelopmental DIsorder Genetics (NEUDIG): a multidisciplinary pathway to new molecular diagnoses by integrating genomic, transcriptomic, and functional analyses. (PRIN 2020,20203P8C3X), Partner (2022-2025) 135 K€

-SURVEIL-Development of a genomic surveillance platform to fight the covid-19 pandemic-(INFRAP2,Finpiemonte Piedmont Region) PI, 174k€

-SARS-CoV-2 and Multiple Sclerosis: Has the Interplay Started? A Study on the Impact of Infection and Vaccine on the Development of Multiple Sclerosis Fondazione Italiana Sclerosi Multipla (2021_Special-Multi_002) Partner (2022-2025) 270 K€

-Fine mapping and functional characterization of MS associated loci to dissect the pathogenetic role of drug target genes Fondazione Italiana Sclerosi Multipla (2019/R/33), Principal Investigator (PI),(2019-2022), 233k€

- Exploring genome/phenome interaction in Multiple Sclerosis (Italian Ministry of Health, RF-2016-02361294), Partner (2018-2021), 90k€

- Identification of genetic risk factors and interaction between genetic and non-genetic risk factors in pediatric multiple sclerosis (PEDiatric Italian Genetic and enviRonment ExposurE) (PEDIGREE study), Fondazione Italiana Sclerosi Multipla (2017/R/15) Partner, 2019-2020, 25k€

-Multiple manifestations of genetic and non-genetic factors in Multiple Sclerosis disentangled with a

multi-omics approach to accelerate personalised medicine. HORIZON 2020 Partner, (2016-2022) ,520K€

-The role of tandem repeats in neurodegenerative diseases: a genomic and proteomic approach MIUR (Italian Ministry of University PRIN 2015), PI, (2016-2020) 146 K€
- Genomic and functional evaluation of the role of the TNFSF14-TNFRSF14 pathway in susceptibility to multiple sclerosis, FISM (Italian Foundation for Multiple Sclerosis) PI, (2016-2019) 185K€
- An integrated genomic, transcriptomic and epigenomic approach in multiplex multiple sclerosis families. Italian Ministry of Health (RF-2011-02350347), Partner (2014-2016) 90€

-Genome-wide analysis of DNA tandem repeats in ALS: the role of Repeat-ome"AriSLA (Fondazione Italiana di ricerca per la SLA – Sclerosi Laterale Amiotrofica), PI (2014-2017), 170€

-Dall'identificazione nella sclerosi multipla di fattori causativi ereditabili e non allo studio delle loro interazioni PROGETTO: MIUR, PRIN 08, Partner (2010-2012) 68K€

-POCEMON (Point-Of-Care MONitoring and Diagnostics for Autoimmune Diseases)Commissione Europea FP7-ICT-2007, partner (2010) 36K€

-Progetto Speciale Immunochip,FISM (Fondazione Italiana per la Sclerosi Multipla), PI, (2010-2012), 200k€

-Ricerca di geni coinvolti nella suscettibilità alla Sclerosi Multipla: studio di replicazione e follow-up di markers genetici identificati in uno studio italiano di associazione caso-controllo su tutto il genoma Fondazione CARIPLO-2010 (N. 2010-0728), PI (2010-2012) 300k€

-Mappaggio fine di loci associati a Sclerosi Multipla nella popolazione dell'Italia continentale: dai marcatori genetici tag identificati in studi GWA alle varianti causali FISM (Fondazione Italiana Sclerosi Multipla), PI (2011-2014), 150k€

Memberships an other Scientific activities

-Member of the "Societa' Italiana Genetica Umana" (SIGU)
-Member of the IMSGC (International Multiple Sclerosis Genetics Consortium) Strategy Group
-Member of the Scientific Committee of FISM (Italian Foundation of Multiple Sclerosis), 2014-2018
-Member of the Editorial Board of "Genes and Immunity"
-Associate Editor of "Cytokine"
-Member of IRCAD (Interdisciplinary Research Center for Autoimmune Diseases, UPO, University, Novara, Italy), and President of IRCAD 2014-2018
- coordinator of the "genomics and transcriptomic" Research Facility (http://caad.uniupo.it/servizi) at the UPO center CAAD (Centro Interdipartimentale "Centro di Eccellenza per la Ricerca traslazionale sulle Malattie
Autoimmuni ed Allergiche")

Teaching activities and PhD supervision

Teaching of Medical Genetics (average 160 hours per academic year since 1996) for a) the following master degree courses: Medicine and Surgery, Biological Sciences, Food health and Environment, b) the following bachelor degree courses: Physiotherapy, Laboratory Technician ,and for the following Specialty Schools: Medical Genetics, Surgery, Physiatry , Pathology, Hospital Pharmacy.

Supervisor of PhD Students attending the UPOe PhD Program “Medical Sciences and “Biotechnology, average 2 Students per academic year in the last 15 years.

Other work experience

(e.g. consultancy if any)

Clinical Assistance Activity

From 2010 she started the coordination of an interdisciplinary medical and laboratory group for the clinical and molecular diagnosis of genetic diseases as collaborator consultant with the AOU Maggiore della Carità of Novara, Novara (Italy) with particular attention to the genetics of neurological diseases

- coordinator of the "genomics and transcriptomic" Research Facility (http://caad.uniupo.it/servizi) at the UPO center CAAD, Novara, Italy (Centro Interdipartimentale "Centro di Eccellenza per la Ricerca traslazionale sulle Malattie)

Administrative role and position responsibility

President of the Degree Course in Medicine and Surgery (since 2016).

Director of the Specialty School of Medical Genetics (since 2022),

Component of the School of Medicine (2017-2024)

Deputy Coordinator of the PhD Program Medical Sciences and Biotechnology (since 2014).

Member of the Board (Giunta) of the School of Medicine (since 2016)

Member of the Board (Giunta) of the Department of Health Sciences (2010-2016)

2010-2016 member of the Technical Commission of Pedagogical Teaching Programming of the Medicine and Surgery Course

Since 2014 Member of the Commission of the Library of the School of Medicine

2014-2020 Delegate of the School of Medicine for the University Job Placement Commission

2016-2020 Delegate of the Rector as representative of the University of Eastern Piedmont in the Regional Group on school-work alternation.

Publications (a selection from the last 10 years)

1: Grassano M, Canosa A, D'Alfonso S, Corrado L, Brodini G, Koumantakis E, Cugnasco P, Manera U, Vasta R, Palumbo F, Mazzini L, Gallone S, Moglia C, Dewan R, Chia R, Ding J, Dalgard C, Gibbs RJ, Scholz S, Calvo A, Traynor B, Chio A. Intermediate HTT CAG repeats worsen disease severity in amyotrophic lateral sclerosis. J Neurol Neurosurg Psychiatry. 2024 Sep 6:jnnp-2024-333998. doi: 10.1136/jnnp-2024-333998. Epub ahead of print. PMID: 39242198.

2. Barizzone N, Leone M, Pizzino A, Kockum I; MultipleMS Consortium; Martinelli- Boneschi F, D'Alfonso S. A Scoping Review on Body Fluid Biomarkers for Prognosis and Disease Activity in Patients with Multiple Sclerosis. J Pers Med. 2022 Aug 31;12(9):1430. doi: 10.3390/jpm12091430. PMID: 36143216; PMCID: PMC9501898.

3: Sorosina M, Barizzone N, Clarelli F, Anand S, Lupoli S, Salvi E, Mangano E, Bordoni R, Roostaei T, Mascia E, Zuccalà M, Vecchio D, Cavalla P, Santoro S, Ferrè L, Zollo A; PROGEMUS; Barlassina C, Cusi D, Martinelli V, Comi G, Leone M, Filippi M, Patsopoulos NA, De Jager PL, De Bellis G,

Esposito F, D'Alfonso S, Martinelli Boneschi F. A multi-step genomic approach prioritized TBKBP1 gene as relevant for multiple sclerosis susceptibility. J Neurol. 2022 Aug;269(8):4510-4522. doi: 10.1007/s00415-022-11109-8. Epub 2022 May 12. Erratum in: J Neurol. 2022 Jun 25;: PMID: 35545683; PMCID: PMC9294010.

4: Grassano M, Calvo A, Moglia C, Sbaiz L, Brunetti M, Barberis M, Casale F, Manera U, Vasta R, Canosa A, D'Alfonso S, Corrado L, Mazzini L, Dalgard C, Karra R, Chia R, Traynor B, Chiò A. Systematic evaluation of genetic mutations in ALS: a population-based study. J Neurol Neurosurg Psychiatry. 2022 Jul 27;93(11):1190–3. doi: 10.1136/jnnp-2022-328931. Epub ahead of print. PMID: 35896380; PMCID: PMC9606529.

5: Tavazzi E, Daberdaku S, Zandonà A, Vasta R, Nefussy B, Lunetta C, Mora G, Mandrioli J, Grisan E, Tarlarini C, Calvo A, Moglia C, Drory V, Gotkine M, Chiò A, Di Camillo B; Piemonte, Valle d'Aosta Register for ALS (PARALS), for the Emilia Romagna Registry for ALS (ERRALS). Predicting functional impairment trajectories in amyotrophic lateral sclerosis: a probabilistic, multifactorial model of disease progression. J Neurol. 2022 Jul;269(7):3858-3878. doi: 10.1007/s00415-022-11022-0. Epub 2022 Mar 10. PMID: 35266043; PMCID: PMC9217910.

6: Faghri F, Brunn F, Dadu A; PARALS consortium; ERRALS consortium; Zucchi E, Martinelli I, Mazzini L, Vasta R, Canosa A, Moglia C, Calvo A, Nalls MA, Campbell RH, Mandrioli J, Traynor BJ, Chiò A. Identifying and predicting amyotrophic lateral sclerosis clinical subgroups: a population-based machine- learning study. Lancet Digit Health. 2022 May;4(5):e359-e369. doi: 10.1016/S2589-7500(21)00274-0. Epub 2022 Mar 24. PMID: 35341712; PMCID: PMC9038712.

7: Perciballi E, Bovio F, Rosati J, Arrigoni F, D'Anzi A, Lattante S, Gelati M, De Marchi F, Lombardi I, Ruotolo G, Forcella M, Mazzini L, D'Alfonso S, Corrado L, Sabatelli M, Conte A, De Gioia L, Martino S, Vescovi AL, Fusi P, Ferrari D. Characterization of the p.L145F and p.S135N Mutations in SOD1: Impact on the Metabolism of Fibroblasts Derived from Amyotrophic Lateral Sclerosis Patients. Antioxidants (Basel). 2022 Apr 22;11(5):815. doi: 10.3390/antiox11050815. PMID: 35624679; PMCID: PMC9137766.

8: Canosa A, Grassano M, Moglia C, Iazzolino B, Peotta L, Gallone S, Brunetti M, Barberis M, Sbaiz L, Palumbo F, Cabras S, Manera U, Vasta R, Traynor B, Corrado L, D'Alfonso S, Mazzini L, Calvo A, Chio A. GBA variants influence cognitive status in amyotrophic lateral sclerosis. J Neurol Neurosurg Psychiatry. 2022 Apr;93(4):453-455. doi: 10.1136/jnnp-2021-327426. Epub 2021 Sep 28. PMID: 34583942; PMCID: PMC8921570.

9: Antona A, Varalda M, Roy K, Favero F, Mazzucco E, Zuccalà M, Leo G, Soggia G, Bettio V, Tosi M, Gaggianesi M, Riva B, Reano S, Genazzani A, Manfredi M, Stassi G, Corà D, D'Alfonso S, Capello D. Dissecting the Mechanism of Action of Spiperone-A Candidate for Drug Repurposing for Colorectal Cancer. Cancers (Basel). 2022 Feb 2;14(3):776. doi: 10.3390/cancers14030776. PMID: 35159043; PMCID: PMC8834219.

10: Ivashynka A, Leone MA, Barizzone N, Cucovici A, Cantello R, Vecchio D, Zuccalà M, Pizzino A, Copetti M, D'Alfonso S, Fontana A. The impact of lifetime coffee and tea loads on Multiple Sclerosis severity. Clin Nutr ESPEN. 2022 Feb;47:199-205. doi: 10.1016/j.clnesp.2021.12.014. Epub 2021 Dec 23. PMID: 35063202.

11: Mascia E, Clarelli F, Zauli A, Guaschino C, Sorosina M, Barizzone N, Basagni C, Santoro S, Ferrè L, Bonfiglio S, Biancolini D, Pozzato M, Guerini FR, Protti A, Liguori M, Moiola L, Vecchio D, Bresolin N, Comi G, Filippi M, Esposito F, D'Alfonso S, Martinelli-Boneschi F. Burden of rare coding variants in an Italian cohort of familial multiple sclerosis. J Neuroimmunol. 2022 Jan 15;362:577760. doi: 10.1016/j.jneuroim.2021.577760. Epub 2021 Nov 5. PMID: 34922125.

12: Clarelli F, Barizzone N, Mangano E, Zuccalà M, Basagni C, Anand S, Sorosina M, Mascia E, Santoro S; PROGEMUS; PROGRESSO; Guerini FR, Virgilio E, Gallo A, Pizzino A, Comi C, Martinelli V, Comi G, De Bellis G, Leone M, Filippi M, Esposito F, Bordoni R, Martinelli Boneschi F, D'Alfonso S. Contribution of Rare and Low-Frequency Variants to Multiple Sclerosis Susceptibility in the Italian Continental Population. Front Genet. 2022 Jan 3;12:800262. doi: 10.3389/fgene.2021.800262. PMID: 35047017; PMCID: PMC8762330.

13: Tondo G, Mazzini L, Caminiti SP, Sarnelli MF, Corrado L, Matheoud R, D'Alfonso S, Cantello R, Sacchetti GM, Perani D, Comi C, De Marchi F. Clinical relevance of single-subject brain metabolism patterns in amyotrophic lateral sclerosis mutation carriers. Neuroimage Clin. 2022;36:103222. doi: 10.1016/j.nicl.2022.103222. Epub 2022 Oct 5. PMID: 36223668; PMCID: PMC9668615.

14: Jiang SH, Mercan S, Papa I, Moldovan M, Walters GD, Koina M, Fadia M, Stanley M, Lea-Henry T, Cook A, Ellyard J, McMorran B, Sundaram M, Thomson R, Canete PF, Hoy W, Hutton H, Srivastava M, McKeon K, de la Rúa Figueroa I, Cervera R, Faria R, D'Alfonso S, Gatto M, Athanasopoulos V, Field M, Mathews J, Cho E, Andrews TD, Kitching AR, Cook MC, Riquelme MA, Bahlo M, Vinuesa CG. Deletions in *VANGL1* are a risk factor for antibody-mediated kidney disease. Cell Rep Med. 2021 Dec 21;2(12):100475. doi: 10.1016/j.xcrm.2021.100475. PMID: 35028616; PMCID: PMC8714939.

15: van Rheenen W, van der Spek RAA, Bakker MK, van Vugt JJFA, Hop PJ, Zwamborn RAJ, de Klein N, Westra HJ, Bakker OB, Deelen P, Shireby G, Hannon E, Moisse M, Baird D, Restuadi R, Dolzhenko E, Dekker AM, Gawor K, Westeneng HJ, Tazelaar GHP, van Eijk KR, Kooyman M, Byrne RP, Doherty M, Heverin M, Al Khleifat A, Iacoangeli A, Shatunov A, Ticozzi N, Cooper-Knock J, Smith BN, Gromicho M, Chandran S, Pal S, Morrison KE, Shaw PJ, Hardy J, Orrell RW, Sendtner M, Meyer T, Başak N, van der Kooi AJ, Ratti A, Fogh I, Gellera C, Lauria G, Corti S, Cereda C, Sproviero D, D'Alfonso S, Sorarù G, Siciliano G, Filosto M, Padovani A, Chiò A, Calvo A, Moglia C, Brunetti M, Canosa A, Grassano M, Beghi E, Pupillo E, Logroscino G, Nefussy B, Osmanovic A, Nordin A, Lerner Y, Zabari M, Gotkine M, Baloh RH, Bell S, Vourc'h P, Corcia P, Couratier P, Millecamps S, Meininger V, Salachas F, Mora Pardina JS, Assialioui A, Rojas-García R, Dion PA, Ross JP, Ludolph AC, Weishaupt JH, Brenner D, Freischmidt A, Bensimon G, Brice A, Durr A, Payan CAM, Saker-Delye S, Wood NW, Topp S, Rademakers R, Tittmann L, Lieb W, Franke A, Ripke S, Braun A, Kraft J, Whiteman DC, Olsen CM, Uitterlinden AG, Hofman A, Rietschel M, Cichon S, Nöthen MM, Amouyel P; SLALOM Consortium; PARALS Consortium; SLAGEN Consortium; SLAP Consortium; Traynor BJ, Singleton AB, Mitne Neto M, Cauchi RJ, Ophoff RA, Wiedau-Pazos M, Lomen-Hoerth C, van Deerlin VM, Grosskreutz J, Roediger A, Gaur N, Jörk A, Barthel T, Theele E, Ilse B, Stubendorff B, Witte OW, Steinbach R, Hübner CA, Graff C, Brylev L, Fominykh V, Demeshonok V, Ataulina A, Rogelj B, Koritnik B, Zidar J, Ravnik-Glavač M, Glavač D, Stević Z, Drory V, Povedano M, Blair IP, Kiernan MC, Benyamin B, Henderson RD, Furlong S, Mathers S, McCombe PA, Needham M, Ngo ST, Nicholson GA, Pamphlett R, Rowe DB, Steyn FJ, Williams KL, Mather KA, Sachdev PS, Henders AK, Wallace L, de Carvalho M, Pinto S, Petri S, Weber M, Rouleau GA, Silani V, Curtis CJ, Breen G, Glass JD, Brown RH Jr, Landers JE, Shaw CE, Andersen PM, Groen EJN, van Es MA, Pasterkamp RJ,

Fan D, Garton FC, McRae AF, Davey Smith G, Gaunt TR, Eberle MA, Mill J, McLaughlin RL, Hardiman O, Kenna KP, Wray NR, Tsai E, Runz H, Franke L, Al-Chalabi A, Van Damme P, van den Berg LH, Veldink JH. Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nat Genet. 2021 Dec;53(12):1636-1648. doi: 10.1038/s41588-021-00973-1. Epub 2021 Dec 6. Erratum in: Nat Genet. 2022 Mar;54(3):361. PMID: 34873335; PMCID: PMC8648564.

16: Pilotto S, Gencarelli J, Bova S, Gerosa L, Baroncini D, Olivotto S, Alfei E, Zaffaroni M, Suppiej A, Cocco E, Trojano M, Amato MP, D'Alfonso S, Martinelli- Boneschi F, Waubant E, Ghezzi A, Bergamaschi R, Pugliatti M. Etiological research in pediatric multiple sclerosis: A tool to assess environmental exposures (PEDiatric Italian Genetic and enviRonment ExposurE Questionnaire). Mult Scler J Exp Transl Clin. 2021 Dec 1;7(4):20552173211059048. doi: 10.1177/20552173211059048. PMID: 34868629; PMCID: PMC8640303.

17: Barizzone N, Cagliani R, Basagni C, Clarelli F, Mendozzi L, Agliardi C, Forni D, Tosi M, Mascia E, Favero F, Corà D, Corrado L, Sorosina M, Esposito F, Zuccalà M, Vecchio D, Liguori M, Comi C, Comi G, Martinelli V, Filippi M, Leone M, Martinelli-Boneschi F, Caputo D, Sironi M, Guerini FR, D'Alfonso S. An Investigation of the Role of Common and Rare Variants in a Large Italian Multiplex Family of Multiple Sclerosis Patients. Genes (Basel). 2021 Oct 13;12(10):1607. doi: 10.3390/genes12101607. PMID: 34681001; PMCID: PMC8535321.

18: 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 Oct 1;78(10):1236-1248. doi: 10.1001/jamaneurol.2021.2598. PMID: 34459874; PMCID: PMC8406220.

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