



FONDAZIONE ITALIANA DI RICERCA PER LA SLA
SCLEROSI LATERALE AMIOTROFICA
ENTE DEL TERZO SETTORE

AriSLA 2024 Conference

Research, development and innovation in ALS

Milan

Hotel Meliá Milan

22nd - 23rd November 2024

www.arisla.org



FONDAZIONE ITALIANA DI RICERCA PER LA SLA
SCLEROSI LATERALE AMIOTROFICA
ENTE DEL TERZO SETTORE

**Fondazione Italiana di Ricerca
per la SLA - Sclerosi Laterale Amiotrofica**

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Welcome to the AriSLA 2024 Conference

We are pleased to come together for this new edition of the AriSLA Scientific Conference, an event eagerly expected by researchers and people with ALS to discuss the state of research on the disease and the latest scientific advances in the field.

In the intense two-day program, researchers will present the results of projects funded by AriSLA and recently completed, demonstrating how crucial it still is to study ALS in all its complexity, through basic, pre-clinical and clinical research. With the contribution of expert speakers, we will delve into key research areas critical both to understanding the progression of the disease and to developing effective therapeutic solutions for patients with ALS.

In this edition, we will highlight the work of young researchers by dedicating two poster sessions to the presentation of their studies. We have also scheduled two panel discussions among various ALS stakeholders to address strategic issues in support of research in this field.

We believe that sharing knowledge through opportunities for dialogue such as this one can foster new collaborations and the achievement of results that can have a concrete impact on people's lives. This is the purpose of our Conference and we are grateful to everyone who has contributed in various ways to making it possible, united by a shared trust in research and a commitment to achieving new milestones to the benefit of patients.

Enjoy the Conference!



Lucia Monaco
President of Fondazione AriSLA



Friday 22nd November

- 08.30 - 09.00 Registration
- 09.00 - 09.30 Opening remarks and institutional greetings
Lucia Monaco - President of Fondazione AriSLA
Representatives of Institutions
- 09.30 - 10.15 **Lectio Magistralis**
Ammar Al-Chalabi, *London*
How do we find new treatments for ALS?

Program

Session I From genetics to mechanisms

Moderators: **Serena Carra**, **Nilo Riva**

- 10.15 - 10.35 **Nicola Ticozzi**, *Milan*
Call 2020 - FG - AZYGOS 2.0 - Autozygosity mapping followed by next generation sequencing in unrelated consanguineous individuals to identify novel ALS-associated genes
- 10.35 - 10.55 **Mauro Cozzolino**, *Rome*
Call 2021 - FG - SWITCHALS - Therapeutic correction of alternative splicing defects in hnRNP A2/B1 as a way to counteract amyotrophic lateral sclerosis associated to FUS
- 10.55 - 11.05 **Valeria Gerbino**, *Rome*
Call 2022 - PG - MotorTBK1 - Cell-autonomous mechanisms of neurodegeneration in TBK1-ALS
- 11.05 - 11.15 General discussion
- 11.15 - 11.45 Coffee break 

Session II Identification of new targets

Moderators: **Emanuele Buratti**, **Nadia D'Ambrosi**

- 11.45 - 11.55 **Elena Ziviani**, *Padua*
Call 2021 - PG - mitoALS - Investigating basal mitophagy in ALS: from flies to human neurons

11.55 - 12.05	Manuela Basso, <i>Trento</i> Call 2021 - PG - SENALS - Premature glial senescence in ALS: untangling the puzzle
12.05 - 12.15	Michela Rigoni, <i>Padua</i> Call 2021 - PG - Breath - Boosting regeneration in ALS motor neurons by targeting the periphery
12.15 - 12.25	Bert Blaauw, <i>Padua</i> Call 2022 - PG - SRXinALS - The role of energy consumption of resting skeletal muscle on hypermetabolism in ALS patients
12.25 - 12.35	General discussion
12.35 - 14.00	Lunch 
14.00 - 15.30	Poster session I

Session III

Preclinical therapeutic approaches

Moderators: **Valentina Bonetto, Antonia Ratti**

15.30 - 15.50	Marta Fumagalli, <i>Milan</i> Call 2019 - FG - GPR17ALS-1 - New strategies to enhance the trophic functions and remyelinating abilities of adult NG2-glia in amyotrophic lateral sclerosis via the GPR17 receptor
15.50 - 16.10	Luca Muzio, <i>Milan</i> Call 2019 - FG - TRAILER - Therapeutic effects of retromer stabilization in amyotrophic lateral sclerosis
16.10 - 16.30	Fabrizio d'Adda di Fagagna, <i>Milan</i> Call 2020 - FG - DDR &ALS - Targeting the DNA damage response to rescue neurodegeneration in ALS
16.30 - 16.40	Savina Apolloni, <i>Rome</i> Call 2021 - PG - ReNicALS - Repurposing niclosamide in ALS
16.40 - 17.00	General discussion
17.00 - 19.00	Poster session II - <i>Wine and cheese</i>

Saturday 23rd November

Session IV From translational to clinical research

Moderators: **Anna Ambrosini, Lucia Monaco**

- 09.00 - 09.45 **Lectio Magistralis**
Giuseppe Lauria, *Milan*
Drug repurposing in ALS
- 09.45 - 10.30 **Round Table**
Valentina Bonetto, Giuseppe Lauria, Annamaria Merico
From translational research to clinical trials
- 10.30 - 11.00 Coffee break ☕
- 11.00 - 11.45 **Round Table**
Anna Ambrosini, Martina de Majo, Fulvia Massimelli
Patient and researchers' synergy for new research strategies
- 11.45 - 12.15 **Lucia Monaco**, *Milan*
The international landscape of rare diseases
- 12.15 - 12.45 AriSLA Call 2024: the funded projects
- 12.45 - 13.00 Poster Award "Giovani per la Ricerca"
Awarded by **Alessandro Marocchi**, "Io Corro con Giovanni"
Association
Closing remarks

Speakers and Moderators of the Meeting

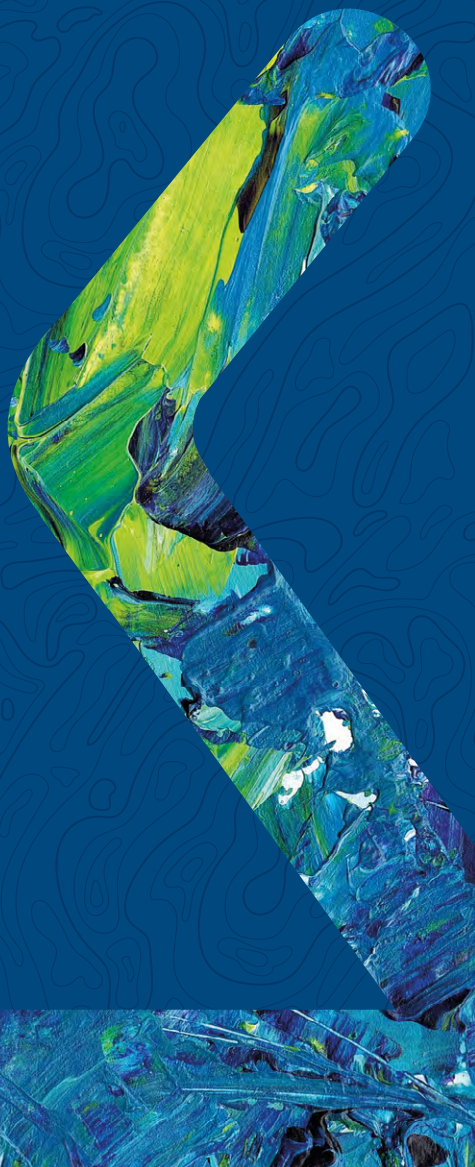


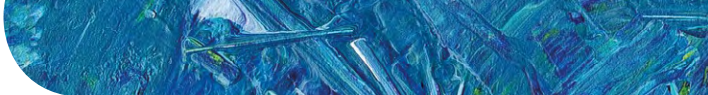
Al-Chalabi Ammar	King's College London, London, UK
Ambrosini Anna	Head of Research, Fondazione AriSLA, Milan
Apolloni Savina	Department of Biology, University of Rome Tor Vergata, Rome
Basso Manuela	Department of Cellular, Computational and Integrative Biology (CIBIO), University of Trento, Trento
Blaauw Bert	Veneto Institute of Molecular Medicine (VIMM), Padua
Bonetto Valentina	Department of Neuroscience, Mario Negri Pharmacological Research Institute IRCCS, Milan
Buratti Emanuele	International Centre for Genetic Engineering and Biotechnology (ICGEB), Trieste
Carra Serena	Department of Biomedical and Metabolic Sciences and Neuroscience, University of Modena and Reggio Emilia, Modena
Cozzolino Mauro	Institute of Translational Pharmacology, National Research Council, Rome
d'Adda di Fagagna Fabrizio	IFOM ETS -The AIRC Institute of Molecular Oncology, Milan
D'Ambrosi Nadia	Department of Biology, University of Rome Tor Vergata, Rome
de Majo Martina	International Alliance of ALS/MND Associations
Fumagalli Marta	Department of Pharmacological and Biomolecular Sciences, University of Milan, Milan
Gerbino Valeria	Santa Lucia IRCCS Foundation, Rome
Lauria Giuseppe	IRCCS Carlo Besta Neurological Institute, University of Milan, Milan
Marocchi Alessandro	'Io Corro Con Giovanni' Association
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Monaco Lucia	President, Fondazione AriSLA, Milan
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Ratti Antonia	IRCCS Italian Auxological Institute, University of Milan, Milan
Rigoni Michela	Department of Biomedical Sciences, University of Padua, Padua
Riva Nilo	IRCCS Carlo Besta Neurological Institute, University of Milan, Milan
Ticozzi Nicola	IRCCS Italian Auxological Institute, University of Milan, Milan
Ziviani Elena	Department of Biology, University of Padua, Padua



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Poster





Poster

P01 Characterization of human i3 motor neurons exposed to CSF from ALS patients stratified by C9orf72 and UNC13A genotype

V. Casiraghi, E. Pellegrini, A. Brusati, S. Peverelli, S. Invernizzi, S. Santangelo, C. Colombrita, V. Silani, A. Ratti

P02 DIG-ALS - Genome wide investigation of oligogenic inheritance in ALS: searching for digenic disease genes with machine learning in a population-based cohort

F. Caushi, B. Bartilomo, M. Grassano, A. Bottrighi, N. Pomella, D. Cerotti, M. Pennisi, A. Sapienza, M. Lai, S. Vilella, F. De Marchi, S. Zucca, F. De Paoli, A. Calvo, R. Vasta, U. Manera, C. Moglia, M. Brunetti, A. Follenzi, P. Bossolasco, L. Corrado, L. Mazzin, A. Ratti, A. Chiò, S. D'Alfonso

P03 One gene, many phenotypes: altered molecular and cellular mechanisms in KIF5A-associated neurodegenerative and neurodevelopmental disorders

M. Cozzi, S. Magri, B. Tedesco, G. Patelli, V. Ferrari, E. Casarotto, M. Chierichetti, P. Pramaggiore, L. Cornaggia, M. Piccolella, M. Galbiati, P. Rusmini, V. Crippa, J. Mandrioli, D. Pareyson, C. Pisciotta, S. D'Arrigo, A. Ratti, L. Nanetti, C. Mariotti, E. Sarto, V. Pensato, C. Gellera, D. Di Bella, R. Cristofani, F. Taroni, A. Poletti

P04 Investigating neurodevelopmental origins and RNA-based therapeutic interventions in C9orf72 ALS patient-derived 3D cortical organoids

D. Franzone, G. Damaggio, M. Paulis, L. Ottoboni, S. Corti, S. Lodato

P05 Defective primary cilia in NEK1 and C9orf72 iPSC-derived motoneurons and brain organoids

S. Invernizzi, S. Santangelo, V. Casiraghi, S. Peverelli, P. Podini, A. Quattrini, V. Silani, P. Bossolasco, A. Ratti

P06 Electrical synapses and hemichannels in ALS models

C. Marullo, S. Zuffetti, L. Croci, S. Taverna, F. Codazzi, G. G. Consalez

P07 Unraveling the role of SUMO2/3 as a modifier of TDP-43 solubility: a new therapeutic avenue for ALS

E. M. Verde, L. Mediani, F. Antoniani, S. Crotti, V. Secco, M. C. Ferrara, A. Rosa, E. Buratti, T. M. Franzmann, S. Alberti, S. Carra

P08 C9orf72 HRE number causes different R-loop distribution and DNA damage signature in ALS patients

W. Sproviero, A. Iacoangeli, A. Jones, A. Al Khleifat, S. Gagliardi, O. Pansarasa, C. Cereda, A. Al-Chalabi, D. Sproviero

P09 NOSRESCUEALS - Unraveling the protective effects of NOS1AP in the rescue of TDP-43 loss of function pathological mechanisms

L. Zangrando, S. Cappelli, A. Spalloni, F. Feiguin, P. Longone, E. Buratti

P10 Endothelial activation in motor neuron disease

G. P. Bhat, S. Diprima, C. Beccaria, P. Podini, A. Motta, I. Brambilla, A. Badaloni, L. Martins, C. Malpighi, G. Nardo, C. Bendotti, N. Riva, A. Quattrini, D. Bonanomi

P11 HDAC6 and TDP-43 promote autophagy impairment in amyotrophic lateral sclerosis

M. Bordoni, E. Scarian, C. Viola, L. Diamanti, F. Dragoni, R. Di Gerlando, B. Rizzo, S. Gagliardi, O. Pansarasa

P12 Translatome profiling reveals deregulated neurovascular crosstalk in amyotrophic lateral sclerosis

I. Brambilla, S. De Pretis, A. Badaloni, C. Malpighi, G. Bhat, D. Bonanomi

P13 The lncRNA ZEB1-AS1 is related to protein FUS and impacts on the deregulation of glutamatergic synapse targets: possible implication in sporadic amyotrophic lateral sclerosis

F. Rey, E. Maghraby, A. Girau, C. Cereda, **S. Carelli**

P14 Genotypically distinct interferon response signatures in ALS brains correlate with disease severity

O. Carletta, O. M. Rifai, C. Perfetto, F. Manganeli, F. Waldron, T. Maniatis, J. Gregory, V. Gerbino

P15 The tetraspanin CD63 facilitates the removal of TDP-43 insoluble species from immortalized motoneuronal cells via extracellular vesicles

E. Casarotto, P. Koshal, A. C. Conti, M. Brodnanovà, M. Chierichetti, L. Cornaggia, M. Cozzi, V. Ferrari, R. F. M. Parra, A. Mohamed, M. Piccolella, P. Pramaggiore, B. Tedesco, R. Cristofani, M. Galbiati, P. Rusmini, R. Cascella, A. Poletti, V. Crippa

P16 SHHield - Targeting smoothened to promote neuroprotection in ALS

M. Ciuro, M. Sangiorgio, V. Caniglia, S. Mastrogiovanni, D. D'Arco, S. Giunta, R. Gulino

P17 Investigating plasma and brain cholesterol esterification in patients with amyotrophic lateral sclerosis

C. Comi, L. Calabresi, C. Pavanello, M. Farè, L. Tremolizzo, F. Gerardi, F. Cerri

P18 Cardiac innervation is compromised by muscle-derived miR-206

V. Di Mauro, M. Ronfini, V. Prando, A. Franco-Romero, M. Albiero, L. Poli, L. Dokshokova, M. Capri, S. Salvioli, C. Franceschi, M. Guescini, M. Sandri, M. Mongillo, T. Zaglia

P19 Unveiling MAP1B as a new possible phenotypic determinant for sporadic bulbar ALS patients

F. Dragoni, R. Di Gerlando, B. Rizzo, L. Diamanti, M. Bordoni, E. Scarian, C. Viola, O. Pansarasa, S. Gagliardi

P20 Acetylation of cyclophilin A reverses TDP-43 mislocalization in PBMCs of ALS patients: potential biomarker and therapeutic target

M. Favagrossa, L. Pasetto, S. Scozzari, S. F. Columbro, G. De Marco, A. Calvo, V. Bonetto

P21 Multi-OMICs approaches to characterize patient-derived spinal cord organoids and assess novel genes associated with C9orf72-amyotrophic lateral sclerosis

N. Galli, M. Rizzuti, L. Ottoboni, J. Ongaro, M. Nizzardo, G. P. Comi, S. Corti

P22 Interfering with FUS protein aggregation with in silico-designed RNA aptamers

M. Gilodi, A. Armaos, J. Rupert, E. Zacco, G. G. Tartaglia

P23 SUMOylation influences TDP-43 aggregation and re-localization

R. Marino, P. Cappelletti, R. Maccarone, M. Corbo, M. Feligioni

P24 FUS orchestrates metabolic processes associated with ALS pathophysiology

J. Awad, P. Cauchy, S. Megat, Q. Raas, L. Dupuis, S. Ciura, E. Kabashi, A. Marian, X. Mingaj

P25 The RNA-binding protein HuD/ELAVL4 as a possible new target in FUS and sporadic ALS

B. Silvestri, M. Mochi, M. Medici, B. Borhy, V. de Turris, A. Colantoni, D. Mawrie, M. G. Garone, C. P. Zammerilla, E. N. Anderson, M. Simula, M. Ballarino, A. Setti, U. B. Pandey, A. Rosa

P26 Generation and characterization of spinal cord organoids for the study of amyotrophic lateral sclerosis

E. Scarian, M. Bordini, C. Viola, F. Dragoni, R. Di Gerlando, B. Rizzo, R. Fontana, L. Diamanti, S. Gagliardi, O. Pansarasa

P27 miR-129-5p: a key factor and therapeutic target in amyotrophic lateral sclerosis

F. Sironi, A. Romano, L. Ottoboni, M. Rizzuti, V. Melzi, D. Gagliardi, F. Verde, A. Ratti, G. P. Comi, N. Ticozzi, V. Silani, S. Corti

P28 Site-directed RNA editing for correcting point mutations causing fALS

D. Capuano, P. Frugis, M. De Robertis, A. Barbon, G. Pesole, E. Picardi, A. M. D'Erchia

P29 A gene therapy approach targeting TDP-43 pathology for ALS (GATTALS)

S. F. Columbro, A. Ghioni, M. Favagrossa, F. Conci, P. V. Fioretti, M. Basso, L. Pasetto, V. Bonetto

P30 Drug X: a repurposable compound mitigating SOD1-ALS phenotypes in fly and murine models

F. Liguori, S. Amadio, I. Passaro, F. Alberti, M. Armida, S. Bullitta, C. Angioli, A. Ferriero, R. L. Potenza, F. Verni, C. Volonté

P31 Enhancing skeletal muscle regeneration in ALS via intramuscular allosteric activation of the P2X7 receptor

C. Margotta, P. Fabbrizio, S. Caravita, A. Passoni, A. Lanno, N. Innocenti, C. Nastasi, A. Tosolini, F. Steyn, E. Davoli, E. Biasini, S. Ngo, G. Nardo

P32 Induced Pluripotent Stem Cell (iPSC)-derived microglia and motor neurons for ALS modelling

E. Pellegrini, C. Lattuada, S. Invernizzi, V. Casiraghi, S. Santangelo, V. Silani, A. Ratti, P. Bossolasco

P33 Targeting upper motor neurons: a cutting-edge neural stem cell therapy for amyotrophic lateral sclerosis

L. Quetti, L. Sali, M. Nizzardo, L. Ottoboni, F. Rizzo, S. Corti, G. P. Comi

P34 A selective ligand targeting the oligodendroglial GPR17 receptor improves functional outcome and myelin integrity in female SOD1G93A mice

S. Raffaele, F. C. Mannella, N. Nguyen, M. Milanese, G. Bonanno, M. P. Abbraccio, T. Bonifacino, M. Fumagalli

P35 MyoMusALS - Exploring the role of the myokine musclin against the progression of amyotrophic lateral sclerosis in mouse models

A. D. Re Cecconi, M. Barone, M. Tortarolo, F. Palo, L. Zentilin, G. Sorarù, G. Nardo, C. Bendotti, R. Piccirillo

P36 Interleukin-9 and its receptor are involved in neuronal activity within murine spinal cord

C. Stacchiotti, S. De Stefano, P. Longone, A. Spalloni, E. Volpe

P37 Neurochemical pathways are altered in ALS

M. Manfredi, T. Chipurura, B. Purghè, L. Mazzini, F. De Marchi, M. Bellan

P38 Characterization of the cellular proliferation and mitochondrial bioenergetic parameters of 5 ALS patient-patient-derived fibroblasts

F. Ciaiola, R. de Mori, M. de Bardi, V. Nesci, G. Borsellino, C. Valle, A. Ferri, R. Massa, L. Boffa, E. Marchionni, A. M. Nardone, M. R. D'Apice, G. Novelli, N. B. Mercuri, E. Buratti, A. Spalloni, P. Longone

P39 Identification of diagnostic and therapeutic ALS targets through cell-type specific analysis of motor nerve damage (IDEALS)

A. Giordano, R. Lombardi, M. Viziello, T. Domi, L. Pozzi, G. Bath, V. Faltracco, M. Cosonni, E. Dalla Bella, A. Quattrini, G. Lauria Pinter, D. Bonanomi, N. Riva

P40 Discovery of biomarkers and TARgetable cerebrospinal fluid protein COMplexes and interactions for ALS diagnosis and therapeutic approaches (TARCO4ALS)

V. De Giorgis, F. Mohammadinezhad, L. Tapella, L. Corrado, L. Mazzini, F. De Marchi, M. Manfredi

P41 Accelerated progression of amyotrophic lateral sclerosis associated with the KIF5A p.Pro986 leu risk variant

A. Manini, R. Vasta, A. Brusati, F. Scheveger, S. Peverelli, A. Maranzano, A. Doretto, F. Gentile, E. Colombo, M. Brunetti, C. Moglia, A. Canosa, U. Manera, M. Grassano, D. Gentilini, S. Messina, F. Verde, C. Morelli, J. E. Landers, B. J. Traynor, A. Chiò, V. Silani, A. Calvo, A. Ratti, N. Ticozzi

P42 Bulbar onset ALS clustering by clinical informed multi-omics (BULB-OMICS)

M. Vizziello, V. Faltracco, A. Giordano, E. Mehmeti, G. Farinazzo, A. De Palma, D. Perico, R. Lombardi, N. Riva, E. Salvi, S. Marcuzzo, M. Consonni, G. Lauria, P. Mauri, E. Dalla Bella

P43 Multimodal assessment of bulbar decline in ALS using audio-visual and machine learning approaches

F. Pierotti, G. Currao, F. Bianchi, B. Fattori, G. Siciliano, [A. Bandini](#)

P44 An emerging era in Italy: the impact of the PALS-led volunteers from Associazione conSLancio Onlus

[M. Consonni](#), SS. Pozzi, R. Piovesana, T. Petrozziello, M. de Majo, M. Peviani, D. Sproviero, M. Boido, S. Gagliardi, F. De Marchi, E. Grossini, J. C. Ayala, A. Desiderato, G. Merlini, R. Schellino, S. Stanga, S. F. Conte, N. De Rossi, A. Zicchieri

P45 Social functioning during lifetime influences the extent of behavioural decline in frontotemporal lobar degeneration

[V. Faltracco](#), A. Telesca, E. M. Giuga, G. Gandini, A. Ferraro, P. Amami, E. Soldini, C. Villa, S. Prioni, S. Piacentini, P. Caroppo, N. Riva, E. Dalla Bella, G. Lauria, M. Consonni

P46 M-AERIALS: Multiparametric assesment of early respiratory involment in ALS - preliminary results

[L. Fontanelli](#), C. Meoni, F. Cucchiara, M. Monfroi, I. Corda, S. Cappelli, L. Becattini, V. Montano, M. Gherardi, F. Bianchi, C. Carlesi, L. Carrozzi, F. Corradi, G. Siciliano

P47 Improving online caregiver training for ALS patients in Italy: a two-year update

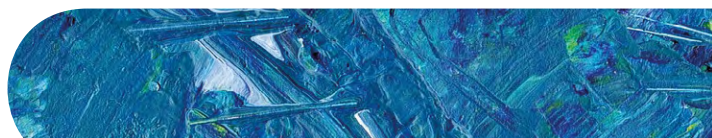
[M. Peviani](#), A. Desiderato, S. F. Conte, S. Pozzi, P. Contessa, J. C. Ayala, M. Consonni, T. Petrozziello, E. Grossini, S. Gagliardi, S. Rota, F. De Marchi, R. Schellino, S. Stanga, G. Merlini, N. De Rossi, A. Zicchieri

P48 EVTestInALS: extracellular vesicles in ALS; testing their use as biomarkers for prognosis and disease progression

L. Donini, P. Gasperini, I. Pesce, R. Frigerio, G. Tomè, M. Pedrotti, P. V. Fioretti, D. Peroni, R. Belli, A. Cutarelli, G. De Marco, I. Merulla, C. Paolantonio, L. Pasetto, T. Tebaldi, F. Rinaldi, A. Ratti, F. Verde, N. Ticozzi, N. Riva, J. Mandrioli, A. Gori, V. Bonetto, R. Zuccarino, R. Tanel, A. Calvo, M. Basso, [L. Pisoni](#)

P49 Emotions, trauma, and quality of life in ALS patients: preliminary results

[E. Zambetti](#), I. Baronchelli, P. Manzoni, C. Mucci, A. Greco



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