CURRICULUM VITAE

TESTI SILVIA

Personal data
Place and date of birth: Zevio, Verona, Italy; 2nd August 1983
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Education

2005 Bachelor Degree in Molecular Biology (score:102/110); University of Padua. Dissertation on "Espression analyses in murine transgenic lines".

2007 Master's Degree in Molecular Biology (score:110/110); University of Padua. Dissertation on "Primary degenerative dementias: mutational analysis of associated genes".

2011 PhD in Molecular, Industrial and Environmental Biotechnologies; University of Verona. Dissertation on "Dissecting the genetic heterogeneity of familial dementias".

Academic position

2011-2013 Research Scholarship Holder at the Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona. Research project: "Diagnostic and prognostic genetic biomarker of Mild Cognitive Impairment". Research manager: Prof. Gian Maria Fabrizi. Project financed by Fondazione Cariverona (grant No. 2009.1026): "Cognitive and behavioral disability in dementias and psychosis" – Scientific coordinator: Prof. Nicolò Rizzuto.

2014-2015 Research Scholarship Holder at the Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona. Research project: "Next generation sequencing in familial forms of neurodegenerative and neuromuscular disorders (Alzheimer's disease, Frontotemporal Lobar Degeneration, Amyotrophic Lateral Sclerosis and Charcot Marie Tooth disease - CMT)". Research manager: Prof. Gian Maria Fabrizi. Project financed by Fondazione Telethon (grant No. GUP13006).

2015-2016 Research Scholarship Holder at the Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona. Research project: "Next generation sequencing in familial forms of neuropathies (Charcot Marie Tooth disease)". Research manager: Prof. Gian Maria Fabrizi. Project financed by ACMT-Rete.

2016-2018 Research Scholarship Holder at the Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona. Research project: "Identification of diagnostic and prognostic genetic biomarkers of neurodegenerative dementias: Mild Cognitive Impairment (MCI), Alzheimer's disease (AD) and, Frontotemporal dementia (FTD) – Amyotrophic Lateral Sclerosis (ALS) spectrum". Research manager: Prof. Gian Maria Fabrizi. Project financed by Fondazione Cariverona (grant No. 2015.0872): "Diagnostic and prognostic biomarkers in

neoplastic, inflammatory and neurodegenerative disorders" - Scientific coordinator: Prof. Giorgio Berton.

2018-2019 Research Scholarship Holder at the Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona. Research project "Molecular bases of lower motor neuron diseases (MND). Next generation sequencing strategies". Research manager: Prof. Gian Maria Fabrizi. Project financed by ACMT-Rete.

2007-present works at the Neuropathology laboratory c\o G.B. Rossi Hospital, AOUI of Verona, where, in addition to research activities she takes care of the molecular diagnosis of some forms of dementias (AD, FTD, CADASIL, Creutzfeldt-Jakob disease, etc.) and neuromuscular disorders (MND, ALS, CMT, etc.).

Acquired Skills

Genetic analyses: isolation and purification of nucleoacids (DNA and RNA) from blood and tissues; DNA and RNA quantification and qualification by Qubit and Bioanalyzer; PCR, gel electrophoresis, **DHPLC** (*Denaturing High Performance Liquid Chromatography*), **Sanger sequencing**; ***Next-Generation Sequencing*** (**NGS**) on **Ion Torrent PGM** (Personal Genome Machine); expression analysis by **RNA-seq** on **Illumina HiSeq1000** and **NextSeq500**; haplotype analysis by **Illumina VeraCode Universal Capture Beads** with **ASPE assay** (Allele-Specific Primer Extension); ELISA; Real Time-PCR; MLPA (Multiplex Ligation-dependent Probe Amplification); tandem repeats expansion analysis by Repeat Primed-PCR and capillary electrophoresis; enzymatic restrictions; genomic subcloning.

Bioinformatic skills: knowledge and use of nomenclature for describing mutations (nucleotide and protein) in accordance with the rules defined by the ***Human Gene Variation Society*** (HGVS); bioinformatic characterization of nucleotide variations [**phylogenetic conservation analyses** (Blastp, CLUSTALW2, etc.); **in silico predictions** of phatogenetic significance of variations (SIFT, Polyphen2, MutationTaster, etc.); **alternative splicing prediction** (NNSplice, GeneSplicer, Human Splicing Finder, etc.)]; **RNA-seq data analysis** by alignment and assembly softwares (e.g.: Bowtie, STAR, SAMTools, TopHat, Cufflinks, HISAT, StringTie) and, transcripts quantification and differential gene expression analysis (DESeq by R, Ballgown); **Gene Ontology** analysis (GO, Kell, CytoScape); **analysis and interpretations of NGS data** from target sequencing by Ion Torrent PGM (Ion Reporter, IGV, SNPs database).
Other expertise: proteins isolation from tissues, western-blot analysis. Submission of research projects to Ethics Committee (EC) of AOUI of Verona (drafting of protocols, informative sheets, informed consent forms and other modules specifically required by EC based on submitted research study type.
Co-author of four research projects [Research manager: Associated Prof. Gian Maria Fabrizi, (disciplinary sector MED/26 – NEUROLOGY) c/o Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona:

- *"Role of Mitochondria in the pathogenesis of Amyotrophic Lateral Sclerosis (ALS): genetical and biochemical evidences, and candidate biomarkers"*, cofounded by Department of Neurosciences, Biomedicine and Movement Sciences, University of Verona – call FUR 2014;

- "*Establishment of a biobank of biological material and associated data, for scientific medical research in the field of neuropathies, dementias and motorneuron diseases*", approved by EC) of AOUI of Verona (Prot. n.: BIOB-NEU-DNA-2014) and funded by AOUI (Indipendent Research Fund, Resolution of the General Menager n.862 of 20/10/2015);

- "*Early biomarkers for neuromuscular damage in Amyotrophic Lateral Sclerosis: a multidisciplinary approach based on muscle and nerve imaging, and skeletal muscle high resolution respirometry and transcriptome profiling*", call PRIN 2015 - positively assessed but not financed;

- "*Validation of a CE-IVD NGS panel for the diagnosis of axonal forms of Charcot-Marie-Tooth disease*", Joint Projects 2016 University of Verona, positively assessed but not financed due to depletion of funds.

Refeering

Referee for Journal of Alzheimer's Disease.

Bibliography

Research indexes updated to 20/06/2018:
h-index Scopus: 6
n. of citations: 260

Peer-reviewed articles

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- Bongianni M, Orrú CD, Groveman BR, Sacchetto L, Fiorini M, Tonoli G, Triva G, Capaldi S, **Testi S**, Ferrari S, Cagnin A, Ladogana A, Poleggi A, Colaizzo E, Tiple D, Vaianella L, Castriciano S, Marchioni D, Hughson AG, Imperiale D, Cattaruzza T, Fabrizi GM, Pocchiari M, Monaco S, Caughey B, Zanusso G. Diagnosis of Human Prion Disease Using Real-Time Quaking-Induced Conversion Testing of Olfactory Mucosa and Cerebrospinal Fluid Samples. *JAMA Neurol.* Published online December 12, 2016.

doi:10.1001/jamaneurol.2016.4614.
- **Testi S**, Tamburin S, Zanette G, Fabrizi GM. Co-Occurrence of the C9ORF72 Expansion and a Novel GRN Mutation in a Family with Alternative Expression of Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *J Alzheimers Dis.* 2015;**44**(1):49-56. doi: 10.3233/JAD-141794.
- van der Zee J, Van Langenhove T, Kovacs GG, Dillen L, Deschamps W, Engelborghs S, Matěj R, Vandenbulcke M, Sieben A, Dermaut B, Smets K, Van Damme P, Merlin C, Laureys A, Van Den Broeck M, Mattheijssens M, Peeters K, Benussi L, Binetti G, Ghidoni R, Borroni B, Padovani A, Archetti S, Pastor P, Razquin C, Ortega-Cubero S, Hernández I, Boada M, Ruiz A, de Mendonça A, Miltenberger-Miltényi G, do Couto FS, Sorbi S, Nacmias B, Bagnoli S, Graff C, Chiang HH, Thonberg H, Perneczky R, Diehl-Schmid J, Alexopoulos P, Frisoni GB, Bonvicini C, Synofzik M, Maetzler W, vom Hagen JM, Schöls L, Haack TB, Strom TM, Prokisch H, Dols-Icardo O, Clarimón J, Lleó A, Santana I, Almeida MR, Santiago B, Heneka MT, Jessen F, Ramirez A, Sanchez-Valle R, Llado A, Gelpi E, Sarafov S, Tournev I, Jordanova A, Parobkova E, Fabrizi GM, **Testi S**, Salmon E, Ströbel T, Santens P, Robberecht W, De Jonghe P, Martin JJ, Cras P, Vandenberghe R, De Deyn PP, Cruts M, Sleegers K, Van Broeckhoven C. Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. *Acta Neuropathol.* 2014 Sep;**128**(3):397-410. doi: 10.1007/s00401-014-1298-7. Epub 2014 Jun 5.
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- van der Zee J, Gijselinck I, Dillen L, Van Langenhove T, Theuns J, Engelborghs S, Philtjens S, Vandenbulcke M, Sleegers K, Sieben A, Bäumer V, Maes G, Corsmit E, Borroni B, Padovani A, Archetti S, Perneczky R, Diehl-Schmid J, de Mendonça A, Miltenberger-Miltenyi G, Pereira S, Pimentel J, Nacmias B, Bagnoli S, Sorbi S, Graff C, Chiang HH, Westerlund M, Sanchez-Valle R, Llado A, Gelpi E, Santana I, Almeida MR, Santiago B, Frisoni G, Zanetti O, Bonvicini C, Synofzik M, Maetzler W, Vom Hagen JM, Schöls L, Heneka MT, Jessen F, Matej R, Parobkova E, Kovacs GG, Ströbel T, Sarafov S, Tournev I, Jordanova A, Danek A, Arzberger T, Fabrizi GM, Testi S, Salmon E, Santens P, Martin JJ, Cras P, Vandenberghe R, De Deyn PP, Cruts M, Van Broeckhoven C, van der Zee J, Gijselinck I, Dillen L, Van Langenhove T, Theuns J, Philtjens S, Sleegers K, Bäumer V, Maes G, Corsmit E, Cruts M, Van Broeckhoven C, van der Zee J, Gijselinck I, Dillen L, Van Langenhove T, Philtjens S, Theuns J, Sleegers K, Bäumer V, Maes G, Cruts M, Van Broeckhoven C, Engelborghs S, De Deyn PP, Cras P, Engelborghs S, De Deyn PP, Vandenbulcke M, Vandenbulcke M, Borroni B, Padovani A, Archetti S, Perneczky R, Diehl-Schmid J, Synofzik M, Maetzler W, Müller Vom Hagen J, Schöls L, Synofzik M, Maetzler W, Müller Vom Hagen J, Schöls L, Heneka MT, Jessen F, Ramirez A, Kurzwelly D, Sachtleben C, Mairer W, de Mendonça A, Miltenberger-Miltenyi G, Pereira S, Firmo C, Pimentel J, Sanchez-Valle R, Llado A, Antonell A, Molinuevo J, Gelpi E, Graff C, Chiang HH, Westerlund M, Graff C, Kinhult Ståhlbom A, Thonberg H, Nennesmo I, Börjesson-Hanson A, Nacmias B, Bagnoli S, Sorbi S, Bessi V, Piaceri I, Santana I, Santiago B, Santana I, Helena Ribeiro M, Rosário Almeida M, Oliveira C, Massano J, Garret C, Pires P, Frisoni G, Zanetti O, Bonvicini C, Sarafov S, Tournev I, Jordanova A, Tournev I, Kovacs GG, Ströbel T, Heneka MT, Jessen F, Ramirez A, Kurzwelly D, Sachtleben C, Mairer W, Jessen F, Matej R, Parobkova E, Danel A, Arzberger T, Maria Fabrizi G, **Testi S**, Ferrari S, Cavallaro T, Salmon E, Santens P, Cras P; European Early-Onset Dementia Consortium. A pan-European study of the C9orf72 repeat associated with FTLD: geographic prevalence, genomic instability, and intermediate repeats. *Hum Mutat.* 2013 Feb;**34**(2):363-73. doi: 10.1002/humu.22244. Epub 2013 Jan 4.
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Books chapters

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- Fabrizi GM, Taioli F, Ferrarini M, Cabrini I, **Testi S**, Cavallaro T, Rizzuto N. Malattia di Charcot-Marie-Tooth. Guida alla diagnosi molecolare. *La Neurologia Italiana.* 2009; n°4: 10-16.