Curriculum Vitae

Personal information

Name	**Massimo Santoro**
Work address	ENEA-CR Casaccia, Via Anguillarese 301, 00123 Rome (Italy)
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Nationality	Italy
Data of birth	09/01/1973

Bibliometric Indicators

h-index=16 (Scopus)
Citation numbers= 886 (Scopus)
Papers= 48
Peer Review (https://publons.com/a/1202758)

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Work Experiences

Dates	**March 2021-Present**
Name of employer	"Health and Environment" Division of Health Protection Technologies, ENEA Casaccia Research Center, Rome, Italy.
Occupational or position held	Researcher.
Main activities and responsabilities	Scientific organization and administrative support of the laboratory. Genetics and molecular biology applied to the characterization of the cellular response to non-ionizing radiations, natural and food-derived biomolecules. Nutrigenomic and nutrigenetic.

Dates	**April 2012-February 2021**
Name of employer	Fondazione Don Carlo Gnocchi, Rome, Italy
Occupation or position held	Researcher.
Main activities and responsibilities	Scientific organization and administrative support of the laboratory. Genetics and molecular biology applied to the rare diseases and the study of the molecular mechanisms underlying several neurological disorders. Management of human and financial resources of the laboratory. Co-author in several projects funded.

Dates	**April 2005 - March 2012**
Name of employer	Department of Neuroscience, UCSC, Policlinico “Gemelli”, Rome, Italy.
Occupation or position held	Researcher.
Main activities and responsibilities	- Research project based on the genotype-phenotype correlation of primary muscle cultures and biopsies from patients with myotonic dystrophy type 1 (DM1) and type 2 (DM2). - Research project based on the analysis of gene expression *RYR1*, *SERCA* and $Ca_V1.1$ in correlation to intracellular calcium signals in myotubes of patients DM1 and DM2. - Research project based on the expression of *INSR* gene in the muscle fibers of type 1 and type 2 in patients with DM1 and DM2 by Laser Capture Microdissection.

Dates	**November 2004 – January 2005**
Name of employer	Departments of Physiology and cell Biology and Biochemistry, Texas Tech University Health Sciences center, Lubbock, Texas.
Occupation or position held	Fellow
Main activities and responsibilities	Development of adenoviral and lentiviral vectors for gene therapy in myocytes.
Dates	**November 2000 – November 2004**
Name of employer	Department of Molecular Cardiology , Fondazione "Maugeri", Pavia, Italy.
Occupation or position held	Researcher.
Main activities and responsibilities	- Research project to develop a mouse "knock-in" model for RyR2 gene (Telethon grant, GP0227Y01). - Molecular Screening by SSCP and DHPLC of several genes in patients with complex arrhythmias and CPVT. - Development of eukaryotic expression vectors and adenoviral vectors for the study of *SCN5A CASQ2* genes in myocytes.
Dates	**September 1999 – September 2000**
Name of employer	Department of Genetic, General and Molecular Biology, University of Naples "Federico II", Italy.
Occupation or position held	Fellow
Main activities and responsibilities	Identification of genomic sequences of fruitless and transformer genes of C. capitata using the screening of cDNA genomic library. Development of new vectors to improve the transformation of germinal line of C. capitata.

Education

Dates	**October 2006 - March 2010**
Name and type of organisation providing education and training	Department of Neuroscience, Catholic University, Rome, Italy
Title of qualification awarded	PhD in Biophysics, Graduate School in Neurosciences
Dates	**July 1999**
Name and type of organisation	Department of Genetic, General and Molecular Biology, University of Naples "Federico II", Italy.
Title of qualification awarded	Degree in biological sciences (110/110 cum lode)
Dates	**February 2001**
Name and type of organisation providing education and training	University of Naples "Federico II", Italy
Title of qualification awarded	Practice the profession of biologist

Training

Dates	**June 2009 – June 2009**
Name and type of organisation providing education and training	Leica Microsystems, Rome, Italy.
Title of qualification awarded	Course confocal microscopy.
Dates	**October 2009 – November 2009**
Name and type of organisation providing education and training	Department of Neuroscience, Catholic University, Rome, Italy
Title of qualification awarded	Safety training of medical and non-medical laboratories-risk chemical and carcinogen.

Personal skills and compentence

Mother tongue	Italian
Other languages	English

Expertise in molecular biology

DNA, RNA and protein extraction.
Southern Blot, Northern Blot, Western Blot.
Molecular cloning.
DNA sequencing. (ABI PRISM 310, Applied Biosystems)
Library screenig (Genomic and cDNA).
Single-Strand Conformation Polymorphism (SSCP).
Denaturing High-Performance Liquid Chromatography (DHPLC).
Polymerase Chain Reaction (PCR), PCR-Real Time and Reverse Transcriptase PCR (RT-PCR), High Resolution Melting, Genotyping, Epigenetics. (StepOnePlus, Applied Biosystems).
Pyrosequencing (PyroMark Q24, Qiagen)
Eucariotic Expression vectors.
Adenoviral vectors for gene therapy.
Cell coltures (HEKA, COS, myoblasts, myotubes and fibroblasts).
RNA-Fluorescence in situ Hybridization (RNA-FISH).
Laser Capture Microdissection (LCM). (Arcturus).
Immunofluorescence (IF) and immunohistochemistry.
Histology (hematoxylin / eosin, ATPasi, DPNH e COX).
Fluorescent and confocal microscopy.

Computer Skills

Google Office.
E-mail e Internet.
Office: Word, Excel e Power point.
Windows (98, 2000, XP, Vista, Windows 7, Windows 10).
Adobe Photoshop e Adobe Illustrator.
GraphPad Prism 8.0

Bioinformatics expertise

DNA Strider
Oligo 4
Genamics Expression
ESE finder
PubMed
High resolution melting software 3.1
Methyl Primer Express v1.0
Chromas Lite
StepOne Software v2.3
Image Lab
Expression Suite software v1.0.3
PyroMarkQ24 software
Primer Express Software v3.0

Scientific publications

- Giordano R, Petersen KK, **Santoro M**, Pazzaglia C, Simonsen O, Valeriani M, Arendt-Nielsen L. Circulating long non-coding RNA signature in knee osteoarthritis patients with postoperative pain one-year after total knee replacement. Scand J Pain. 2021 Jul 29. doi: 10.1515/sjpain-2021-0069.

- Siotto M, Germanotta M, **Santoro M**, Cipollini V, Guardati G, Papadopoulou D, Bray E, Mastrorosa A, Aprile I. Serotonin Levels and Cognitive Recovery in Patients with Subacute Stroke after Rehabilitation Treatment. Brain Sci. 2021;11(5):642.

- **Santoro M**, Siotto M, Germanotta M, Mastrorosa A, Papadopoulou D, Aprile I. Association study of SLC6A4 (5-HTTLPR) polymorphism and its promoter methylation with rehabilitation outcome in patients with subacute stroke. Genes. 2021;12(4):579.

- Nociti V, **Santoro M**. What do we know about the role of lncRNAs in multiple sclerosis? Neural Regen Res. 2021;16(9):1715-1722.

- Petrillo S, **Santoro M**, La Rosa P, Perna A, Gallo MG, Bertini ES, Silvestri G, Piemonte F. Nuclear Factor Erythroid 2-Related Factor 2 Activation Might Mitigate Clinical Symptoms in Friedreich's Ataxia: Clues of an "Out-Brain Origin" of the Disease From a Family Study. Front Neurosci. 2021;15:638810.

- **Santoro M**, Siotto M, Germanotta M, Bray E, Mastrorosa A, Galli C, Papadopoulou D, Aprile I. Bdnf rs6265 polymorphism and its methylation in patients with stroke undergoing rehabilitation. International Journal of Molecular Sciences, Volume 21, Issue 22, 2 November 2020, Article number 8438, Pages 1-11.

- Siotto M, Germanotta M, **Santoro M**, Di Blasi C, Loreti C, Mastropaolo S, Aprile I. Total serum calcium and recovery after rehabilitation in patients with stroke. Applied Sciences, Volume 10, Issue 21, 1 November 2020, Article number 7893, Pages 1-8.

- **Santoro M**, Perna A, La Rosa P, Petrillo S, Piemonte F, Rossi S, Riso V, Nicoletti TF, Modoni A, Pomponi MG, Chiurazzi P, Silvestri G. Compound heterozygosity for an expanded (GAA) and a (GAAGGA) repeat at FXN locus: from a diagnostic pitfall to potential clues to the pathogenesis of Friedreich ataxia. Neurogenetics, 2020; Pages 279-287.

- **Santoro M**, Piacentini R, Perna A, Pisano E, Severino A, Modoni A, Grassi, C, Silvestri G. Resveratrol corrects aberrant splicing of RYR1 pre-mRNA and Ca2+ signal in myotonic dystrophy type 1 myotubes. Neural Regeneration Research, Volume 15, Issue 9, 1 September 2020, Pages 1757-1766.

- Centofanti F, **Santoro M**, Marini M, Visconti VV, Rinaldi AM, Celi M, D'Arcangelo G, Novelli G, Orlandi A, Tancredi V, Tarantino U, Botta A. Identification of aberrantly-expressed long non-coding rnas in osteoblastic cells from osteoporotic patients. Biomedicines, Volume 8, Issue 3, 1 March 2020, Article number 65.

- Siotto M, **Santoro M**, Aprile I. Vitamin D and rehabilitation after stroke: Status of art. Applied Sciences, Volume 10, Issue 6, 1 March 2020, Article number 1973.

- **Santoro M**, Vollono C, Pazzaglia C, Di Sipio E, Giordano R, Padua L., Arendt-Nielsen L, Valeriani M. ZNRD1-AS and RP11-819C21.1 long non-coding RNA changes following painful laser stimulation correlate with laser-evoked potential amplitude and habituation in healthy subjects: A pilot study. European Journal of Pain Volume 24, Issue 3, 1 March 2020, Pages 593-603.

- Fontana L, **Santoro M**, D'Apice MR, Peluso F, Gori G, Morrone A, Novelli G, Dosa L., Botta A. Identification, molecular characterization and segregation analysis of a variant DMPK pre-mutation allele in a three-generation Italian family. Acta Myologica. Volume 39, Issue 1, 2020, Pages 13-18.

- **Santoro M**, Nociti V, Lucchini M, Loiodice M, Centofanti F, Botta A, Losavio FA, De Fino C, Mirabella M. A pilot study of lncRNAs expression profile in serum of progressive multiple sclerosis patients. Eur Rev Med Pharmacol Sci 2020; 24 (6): 3267-3273.

- Nociti V, **Santoro M**, Quaranta D, Losavio FA, De Fino C, Giordano R, Palomba N, Rossini PM, Guerini FR, Clerici M, Caputo D, Mirabella M. BDNF rs6265 polymorphism methylation in Multiple Sclerosis: A possible marker of disease progression. PLoS One. 2018 Oct 23;13(10):e0206140.

- Signorile PG, Severino A, **Santoro M**, Spyrou M, Viceconte R, Baldi A. Methylation analysis of HOXA10 regulatory elements in patients with endometriosis. BMC Res Notes. 2018 Oct 11;11(1):722.

- Rossi S, Romano A, Modoni A, Perna F, Rizzo V, **Santoro M**, Monforte M, Pieroni M, Luigetti M, Pomponi MG, Silvestri G. Dysautonomia as Onset Symptom of Myotonic Dystrophy Type 2. Eur Neurol. 2018 Mar 13;79(3-4):166-170.

- **Santoro M**, Fontana L, Maiorca F, Centofanti F, Massa R, Silvestri G, Novelli G, Botta A. Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochim Biophys Acta. 2018 Mar;1864(3):917-924.

- Siotto M, Aprile I , Simonelli I, Pazzaglia C, Ventriglia M, **Santoro M**, Imbimbo I, Squitti R, Padua L. An exploratory study of BDNF and oxidative stress marker alterations in subacute and chronic stroke patients affected by neuropathic pain. J Neural Transm (Vienna). 2017 Dec;124(12):1557-1566.

- Maltese M, Martella G, Imbriani P, Schuermans J, Billion K, Sciamanna G, Farook F, Ponterio G, Tassone A, **Santoro M**, Bonsi P, Pisani A, Goodchild RE. Abnormal striatal plasticity in a DYT11/SGCE myoclonus dystonia mouse model is reversed by adenosine A2A receptor inhibition. Neurobiol Dis. 2017;108:128-139.

- Imbriani P, Sciamanna G, **Santoro M**, Schirinzi T, Pisani A. Promising rodent models in Parkinson's disease. Parkinsonism Relat Disord. 2017. pii: S1353-8020(17)30274-2.

- **Santoro M**, Mirabella M, De Fino C, Bianco A, Lucchini M, Losavio F, Sabino A, Nociti V. Sativex® effects on promotermethylation and on CNR1/CNR2 expression in peripheral blood mononuclear cells of progressive multiple sclerosis patients. Journal of the Neurological Sciences 379 (2017) 298–303.

- Di Nottia M, Masciullo M, Verrigni D, Petrillo S, Modoni A, Rizzo V, Di Giuda D, Rizza T, Niceta M, Torraco A, Bianchi M, **Santoro M**, Bentivoglio AR, Bertini E, Piemonte F, Carrozzo R, Silvestri G. DJ-1 modulates mitochondrial response to oxidative stress: clues from a novel diagnosis of PARK7. Clin Genet. 2017 Jul;92(1):18-25.

- **Santoro M**, Masciullo M, Silvestri G, Novelli G, Botta A. Myotonic dystrophy type 1: role of CCG, CTC and CGG interruptions within DMPK alleles in the pathogenesis and molecular diagnosis. Clin Genet. 2016 Dec 19.

- Perfetti A, Greco S, Cardani R, Fossati B, Cuomo G, Valaperta R, Ambrogi F, Cortese A, Botta A, Mignarri A, **Santoro M**, Gaetano C, Costa E, Dotti MT, Silvestri G, Massa R, Meola G, Martelli F. Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. Sci Rep. 2016 Dec 1;6:38174.

- **Santoro M**, Nociti V, Lucchini M, De Fino C, Losavio FA, Mirabella M. Expression Profile of Long Non-Coding RNAs in Serum of Patients with Multiple Sclerosis. J Mol Neurosci. 2016 Mar 31.

- **Santoro M**, Nociti V, De Fino C, Caprara A, Giordano R, Palomba N, Losavio F, Marra C, Patanella AK, Mirabella M, Gainotti G, Quaranta D. Depression in multiple sclerosis: effect of brain derived neurotrophic factor Val66Met polymorphism and disease perception. Eur J Neurol. 2016 Mar;23(3):630-40. doi: 10.1111/ene.12913. Epub 2016 Jan 12.

- Bianchi ML, Leoncini E, Masciullo M, Modoni A, Gadalla SM, Massa R, Rastelli E, Terracciano C, Antonini G, Bucci E, Petrucci A, Costanzi S, **Santoro M**, Boccia S, Silvestri G. Increased risk of tumor in DM1 is not related to exposure to common lifestyle risk factors. J Neurol. 2016 Mar;263(3):492-8.

- **Santoro M**, Fontana L, Masciullo M, Bianchi ML, Rossi S, Leoncini E, Novelli G, Botta A, Silvestri G. Expansion size and presence of CCG/CTC/CGG sequence interruptions in the expanded CTG array are independently associated to hypermethylation at the DMPK locus in myotonic dystrophy type 1 (DM1). Biochim

Biophys Acta. 2015 Dec;1852(12):2645-52.

- Zampetti A, Silvestri G, Manco S, Khamis K, Masciullo M, Bianchi ML, Damiani A, Santoro M, Linder D, Bewley A, Feliciani C. Dysplastic nevi, cutaneous melanoma, and other skin neoplasms in patients with myotonic dystrophy type 1: A cross-sectional study. J Am Acad Dermatol. 2015 Jan;72(1):85-91.

- Spitalieri P, Talarico RV, Botta A, Murdocca M, D'Apice MR, Orlandi A, Giardina E, **Santoro M**, Brancati F, Novelli G, Sangiuolo F. Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. Cell Reprogram. 2015 Aug;17(4):275-87.

- Colamartino M, **Santoro M**, Duranti G, Sabatini S, Ceci R, Testa A, Padua L, Cozzi R. Evaluation of Levodopa and Carbidopa Antioxidant Activity in Normal Human Lymphocytes In Vitro: Implication for Oxidative Stress in Parkinson's Disease. Neurotox Res. 2014 Oct 30

- Bianchi ML, Losurdo A, Di Blasi C, **Santoro M**, Masciullo M, Conte G, Valenza V, Damiani A, Della Marca G, Silvestri G. Prevalence and clinical correlates of sleep disordered breathing in myotonic dystrophy types 1 and 2. Sleep Breath. 2013 Dec 6.

- **Santoro M**, Piacentini R, Masciullo M, Bianchi ML, Modoni A, Podda MV, Ricci E, Silvestri G, Grassi C. Alternative splicing alterations of Ca2+ handling genes are associated with Ca2+ signal dysregulation in DM1 and DM2 myotubes. Neuropathol Appl Neurobiol. 2013 Jul 29.

- **Santoro M**, Masciullo M, Bonvissuto D, Bianchi ML, Michetti F, Silvestri G. Alternative splicing of human insulin receptor gene (INSR) in type I and type II skeletal muscle fibers of patients with myotonic dystrophy type 1 and type 2. Mol Cell Biochem. 2013 Aug;380(1-2):259-65.

- Masciullo M, Iannaccone E, Bianchi ML, **Santoro M**, Conte G, Modoni A, Monforte M, Tasca G, Laschena F, Ricci E, Silvestri G. Myotonic dystrophy type 1 and de novo FSHD mutation double trouble: a clinical and muscle MRI study. Neuromuscul Disord. 2013 May;23(5):427-31.

- **Santoro M**, Masciullo M, Pietrobono R, Conte G, Modoni A, Bianchi ML, Rizzo V, Pomponi MG, Tasca G, Neri G, Silvestri G. Molecular, clinical, and muscle studies in myotonic dystrophy type 1 (DM1) associated with novel variant CCG expansions. J Neurol. 2013 May;260(5):1245-57.

- Masciullo M, **Santoro M**, Modoni A, Ricci E, Guitton J, Tonali P, Silvestri G. Substrate reduction therapy with miglustat in chronic GM2 gangliosidosis type Sandhoff: results of a 3-year follow-up. J Inherit Metab Dis. 2010 Sep 4.

- **Santoro M**, Modoni A, Masciullo M, Gidaro T, Broccolini A, Ricci E, Tonali PA, Silvestri G. Analysis of MTMR1 expression and correlation with muscle pathological features in juvenile/adult onset myotonic dystrophy type 1 (DM1) and in myotonic dystrophy type 2 (DM2). Experimental and Molecular Pathology. 2010;89:158-168.

- Silvestri G, Masciullo M, Piane M, Savio C, Modoni A, **Santoro M**, Chessa L. Homozygosity for c 6325T>G transition in the ATM gene causes an atypical, late-onset variant form of ataxia-telangiectasia. J Neurol. 2010;10:1738-1740.

- Masciullo M, Modoni A, Fattori F, Santoro M, Denora PS, Tonali P, Santorelli FM, Silvestri G.A novel mutation in the SACS gene associated with a complicated form of spastic ataxia. J Neurol. 2008;9:1429-1431.

- **Santoro M**, Modoni A, Sabatelli M, Madia F, Piemonte F, Tozzi G, Ricci E, Tonali PA, Silvestri G. Chronic GM2 gangliosidosis type Sandhoff associated with a novel missense HEXB gene mutation causing a double pathogenic effect. Mol Genet Metab. 2007;91:111-114.

- Modoni A, Contarino MF, Bentivoglio AR, Tavolacci E, **Santoro M**, Calcagni ML, Tonali PA, Neri G, Silvestri G. Prevalence of spinocerebellar ataxia type 2 mutation among Italian Parkinsonian patients. Movement Disorders. 2007;22:324-327.

- Terentyev D, Nori, A, **Santoro M**, Viatchenko-Karpinski S, Kubalova, Z, Gyorke I, Terentyeva R, Vedamoorthyrao S, Blom NA, Valle G, Napoletano C, Williams SC, Volpe P, Priori SG, Gyorke S. Abnormal Interactions of Calsequestrin With the Ryanodine Receptor Calcium Release Channel Complex Linked to Exercise-Induced Sudden Cardiac Death. Circ Res. 2006 ;98:1151-1158.

- Cerrone M*, Colombi B*, **Santoro M***, di Barletta MR, Scelsi M, Villani L, Napolitano C, Priori SG. Bidirectional ventricular tachycardia and fibrillation elicited in a knock-in mouse model carrier of a mutation in the cardiac ryanodine receptor. Circ Res. 2005;96:e77-82. *These authors have contributed equally to this work.

Book

- **Santoro M**, Masciullo M, Novelli G, Botta A. 2015 Variant interrupted DMPK alleles; implication in pathogenesis and molecular diagnosis of myotonic dystrophy type 1 (DM1), chapter 8 pp. 121-137, in Myotonic Dystrophies, epidemiology, diagnosis and therapeutic challenges. Edited by Sandra Jenkins, Nova Biomedica Publishers Inc, New York, USA.

- Saccone G, Pane A, Testa G, **Santoro M**, et al., 2000 Sex determination in medfly: a molecular approach, pp. 491–496 in Area-Wide Control of Fruitflies and Other Pest Insects, edited by K.-H. TAN. Penerbit USM, Penang.

Consento il trattamento dei dati personali ai sensi dell'art. 7 del D.Lgs n.196 del 30.6.2003.