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Prof. Jessica Diana Rosati is Associate Professor of Applied Biology at Saint Camillus International University of Health Sciences.
She is a molecular and cellular biologist, after her PhD in Genetics and Molecular Biology, has expanded her higher field of expertise by pursuing a Specialization in Medical Genetics to have a broader view of all genetic diseases, not only from a molecular/cellular perspective, but also from a genetic perspective.
Since 2017, she has been head of the Cellular Reprogramming Unit at IRCCS Casa Sollievo della Sofferenza Foundation. In recent years, she has studied the molecular mechanisms of rare genetic diseases, publishing numerous scientific papers regarding the production of induced pluripotent human stem cells (iPSCs) from patients affected by diseases such as Huntington's disease, Joubert's syndrome, Smith-Magenis syndrome, Duchenne dystrophy and others. In recent years, the research group she leads has been working on the characterization of molecular pathways implicated in these genetic disorders. Prof. Rosati has optimized a protocol for the neuralization of induced pluripotent stem cells, obtained directly from patient fibroblasts, using an original selection method. This protocol is valid for GMP certification. Among articles published in recent years, she considers this method, a starting point for the study of genetically based neural developmental and neurodegenerative diseases. She is a referee for numerous international scientific journals and collaborates on several international research projects, including as an associate editor. The studies and research coordinated by Prof. Rosati are based on a multidimensional and systemic approach that integrates knowledge and methodologies from multiple disciplinary fields, overcoming traditional reductionist and deterministic approaches.

CURRENT POSITIONS

2024	Associate Professor of Applied Biology, Departmental Faculty of Medicine, Saint Camillus International University of Health Sciences
2017 to date	Head of Cellular Reprogramming Unit, Fondazione IRCCS Casa Sollievo della Sofferenza -Mendel Institute, Rome, Italy

PAST POSITIONS

2015-2016	Senior scientist, Cellular Reprogramming Unit, Casa Sollievo della Sofferenza - Mendel Institute, Rome, Italy
2012-2014	Contract Researcher, Neurogenetics Unit, CSS-Mendel, Roma
2007-2011	Contract Researcher, Vascular Pathology Lab, Istituto Dermopatico dell'Immacolata, Roma
2004-2006	Post-Doc Fellowship at Nucleic Acid Center, CNR c/o Department of Genetics and Molecular Biology, University of Rome "La Sapienza".
2000-2003	PhD student in Genetics and Molecular Biology, University of Rome "La Sapienza"
1998-2000	Training at Nucleic Acid Center (Dr Nasi), CNR c/o Department of Genetics and Molecular Biology, University of Rome "La Sapienza".
1995-1997	Training at the Department of Cellular Biology, University of Rome, "La Sapienza"

MAIN ACADEMIC DEGREES AND QUALIFICATIONS

2024	Associate Professor of Applied Biology, Departmental Faculty of Medicine, Saint Camillus International University of Health Sciences
2018	Awarded title of Associate Professor (National Scientific Qualification - Ministry of Education, Universities and Research (MIUR)- Applied Biology
2017	Specialization in Medical Genetics (60/60 *cum laude*)
2011	Eligibility to the role of ISS Researcher
2011	Eligibility to the role of CNR Researcher
2004	PhD in Genetics and Molecular Biology
2003	Board in Biology
1997	Degree certificate in Biological Sciences (110/110 *cum laude*)

RESEARCH ACTIVITIES AND GRANTS

Year	Title of Research Project	Role	Institute(s)/ Organizations involved
2024-2026	PNRR-POC-2023-12377196: Therapeutic development (TRL4) of muscle speci2c microRNAs for Kennedy's disease.	Principal Collaborator	Italian Ministry of Health
2024-2026	PNRR-MCNT1-2023-12377520: Omic profile in autism spectrum disorder: from cellular level towards future treatments.	Collaborator	Italian Ministry of Health
2023-2026	RF-2021-12372766: Dissemination of ALS neuronal damage through neural circuits and neuroglial interactions: from systems biology to neuroprotection.	Collaborator	Italian Mininstry of Health
2023-2025	*PNRR-MAD-2022-12376068:* Pathogenic role of myelin loss in focal seizures and epilepsy: an integrated approach from neurons to patients	Principal Collaborator	Italian Mininstry of Health
2022-2024	*Institution of a laboratory for the production and characterization of human cell models necessary for the study of rare genetic diseases.*	Scientific Coordination	Fondazione Prosolidar
2021-2024	Una nuova trasmissione su RAI1.	Principal Collaborator	Just Italia Foundation
2020-2021	Architecture of cell differentiation, stress-mediated protein expression and transport in iPSC-derived motor neurons bearing a pG376D TDP-43 mutation.	Principal Collaborator	ARISLA
2019-2021	D-Rhythm. The role of Vitamin D and circadian alterations in neurodegenerative diseases: a clinical and biological study on Parkinson's and Alzheimer's diseases	Principal Collaborator	Alberto Sordi Foundation

Year	Title of Research Project	Role	Institute(s)/ Organizations involved
2018-2020	The roles of biological clock deregulation and retinoic acid signalling impairment in Smith-Magenis syndrome	PI	Jerome Lejeune Foundation
2017-2020	Advanced in vivo and in vitro technologies to STudy Juvenile Huntington Disease neuronal connectivity and its relationship with clinical and genetic factors. The RAREST-JHD project.	CO-PI	Italian Ministry of Health - Finalizzata
2014-2017	Production and characterization of induced pluripotent stem cells from somatic cells of patients affected by genetic and neurodegenerative diseases".	PI	Italian Ministry of Health-Ricerca Corrente

COURSES

Year	Title and Organization
2024	Advanced Project Management Course.
2023	Basic Project Management Course
2015	Training course on "*WT Plus assay and miRNA Flash Tag assay*" organized by Affymetrix
2013	Training course on Human iPSCs Derivation and Culture" organized by University of Cambridge, MRC Centre for Stem Cell Biology and Regenerative Medicine
2011	*Stem Cell Differentiation Training Course*" organized by Stem Cell Fate Lab in collaboration with Euroclone
2005	*Application of bioinformatics to molecular and structural biology*" organized by Dip. of Biochemistry, University of Rome
2004	*Gene Expression Analysis course*" organized from AB.EL Science Ware
1999	*Antibody phage display library* EMBL course, Maastricht, The Netherlands
1998	*Development of new vectors and transcriptional switches for the gene therapy of human disease* course organized by the Universities of Trieste and Udine, and by S.I.B.B.M and CE.PRO.BI.MOL, Cividale del Friuli (UD), Italy
1997	*Molecular mechanism of embryonic development* course organized by the Universities of Trieste and Udine, and by S.I.B.B.M and CE.PRO.BI.MOL, Cividale del Friuli (UD), Italy

PUBLICATIONS: PAPERS AND BOOK CHAPTERS

Date	Title	Publisher
2024	Investigating the impact of the Parkinson's-associated GBA1 E326K mutation on GCase dimerization and interactome dynamics through an in silico approach	Under review

Date	Title	Publisher
2024	TDP-43G376D mutation induces mitochondrial dysfunctionality and energy metabolism rearrangements in fibroblasts from symptomatic and asymptomatic members of an Amyotrophic Lateral Sclerosis family	Under review
2024	Generation of the CSSi020-A (14437) iPSC line from a patient carrying a copy number variation (CNV) in the 17p11.2 chromosome region.	Stem Cell Res. 2024 Sep 4;81:103544. doi: 10.1016/j.scr.2024.103544.
2024	Induced pluripotent stem cell production (CSSi019-A)(14432) from an asymptomatic subject carrying a expansion of C9orf72 gene	Stem Cell Res. 2024 Aug 22;81:103540. doi: 10.1016/j.scr.2024.103540
2024	Clarifying main nutritional aspects and resting energy expenditure in children with Smith-Magenis Syndrome	Eur J Pediatr. 2024 Aug 20. doi: 10.1007/s00431-024-05715-z.
2024	*CircHTT(2,3,4,5,6)* – co-evolving with the *HTT* CAG-repeat tract - modulates Huntington`s Disease phenotypes	Mol Ther Nucleic Acids. 2024 Jun 3;35(3):102234. doi: 10.1016/j.omtn.2024.102234.
2024	Production of an induced pluripotent stem cell line CSSi018-A (14192) from a patient with hypomyelinating leukodystrophy 7 (HLD7) carrying biallelic variants of POLR3A (c.1802 T > A; c.4072G > A)	Stem Cell Res. 2024 Aug;78:103468. doi: 10.1016/j.scr.2024.103468.
2024	Amniotic fluid stem cell derived extracellular vesicles educate type 2 conventional dendritic cells to rescue autoimmune disorders.	J Extracell Vesicles. 2024 Jun;13(6):e12446. doi: 10.1002/jev2.12446.
2024	Generation of induced pluripotent stem cells (CSSi017-A)(12862) from a patient carrying a repeat expansion in the C9orf72 gene and with amyotrophic lateral sclerosis.	Stem Cell Res. 2024 Jun;77:103412. doi: 10.1016/j.scr.2024.103412.
2023	Metabolic Profile of Patients with Smith-Magenis Syndrome: An Observational Study with Literature Review	Children (Basel). 2023 Aug 25;10(9):1451. doi: 10.3390/children10091451.
2023	Circadian profile, daytime activity, and the Parkinson's phenotype: A motion sensor pilot study with neurobiological underpinnings	Neurobiol Sleep Circadian Rhythms. 2023 Mar 26;14:100094. doi:10.1016/j.nbscr.2023.100094.
2023	Deepening the understanding of CNVs on chromosome 15q11-13 by using hiPSCs: An overview	Front Cell Dev Biol. 2023 Jan 6;10:1107881. doi: 10.3389/fcell.2022.1107881.
2023	Generation of an induced pluripotent stem cell line CSSi015-A (9553), carrying a point mutation c.2915C > T in the human calcium sensing receptor (CasR) gene	Stem Cell Res. 2023 Jan 7;67:103023. doi: 10.1016/j.scr.2023.103023.
2022	Retinoic acid-induced 1 gene haploinsufficiency alters lipid metabolism and causes autophagy defects in Smith-Magenis syndrome	Cell Death Dis. 2022 Nov 21;13(11):981. doi: 10.1038/s41419-022-05410-7.
2022	Generation and characterization of CSSi016-A (9938) human pluripotent stem cell line carrying two biallelic variants in MTMR5/SBF1 gene resulting in a case of severe CMT4B3	Stem Cell Res. 2022 Dec; 65:102946. doi: 10.1016/j.scr.2022.102946.
2022	Generation of an induced pluripotent stem cells line, CSSi014-A 9407, carrying the variant c.479C>T in the human iduronate 2-sulfatase (hIDS) gene	Stem Cell Res. 2022 Aug;63:102846. doi: 10.1016/j.scr.2022.102846.
2022	Skeletal muscle in polyglutamine diseases: More than a bystander to central nervous system degeneration	Cells. 2022 Jul 3;11(13):2105. doi: 10.3390/cells11132105.
2022	Production of CSSi013-A (9360) iPSC line from an asymptomatic subject carrying an heterozygous mutation in TDP-43 protein	Stem Cell Res. 2022 Aug;63:102835. doi: 10.1016/j.scr.2022.102835.

Date	Title	Publisher
2022	Circadian profile, daytime activity, and the Parkinson's phenotype: A motion sensor pilot study with neurobiological underpinnings	Neurobiol Sleep Circadian Rhythms. 2023 Mar 26;14:100094. doi: 10.1016/j.nbscr.2023.100094.
2022	Smith Magenis syndrome: First case of congenital heart defect in a patient with Rai1 mutation.	Am J Med Genet A. 2022 Apr 4. doi: 10.1002/ajmg.a.62740. PMID: 35373511.
2022	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.
2021	Generation of an induced pluripotent stem cell line (CSS012-A (7672)) carrying the p.G376D heterozygous mutation in the TARDBP protein	Stem Cell Res. 2021 May;53:102356. doi: 10.1016/j.scr.2021.102356. PMID: 34087986.
2021	COVID-19 Specific Immune Markers Revealed by Single Cell Phenotypic Profiling.	Biomedicines 2021 Nov 29;9(12):1794. doi: 10.3390/biomedicines9121794. PMID: 34944610.
2021	Known Drugs Identified by Structure-Based Virtual Screening Are Able to Bind Sigma-1 Receptor and Increase Growth of Huntington Disease Patient-Derived Cells.	Int J Mol Sci. 2021 Jan 28;22(3):1293. doi: 10.3390/ijms22031293.
2021	FUNCTIONAL OUTCOMES OF COPY NUMBER VARIATION OF CHRNA7: CURRENT KNOWLEDGE AND NEW INSIGHTS FROM INDUCED PLURIPOTENT STEM CELLS STUDIES	Advances in Stem Cell Biology - Current Progress in iPSC Disease Modeling. Vol 15, ISBN: 978-0-12-823882-0.
2021	Smith-Magenis Syndrome: From genetics to clinical presentation, disease pathogenesis and model systems.	Advances in Stem Cell Biology - Current Progress in iPSC Disease Modeling. Vol 14, ISBN: 978-0-323-85765-9.
2020	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellis-van Creveld syndrome caused by hypomorphic mutations in the EVC gene.	Hum Mutat. 2020 Dec;41(12):2087-2093. doi: 10.1002/humu.24112.
2020	A Link between Genetic Disorders and Cellular Impairment, Using Human Induced Pluripotent Stem Cells to Reveal the Functional Consequences of Copy Number Variations in the Central Nervous System-A Close Look at Chromosome 15.	Int J Mol Sci. 2020 Mar 9;21(5). pii: E1860. doi: 10.3390/ijms21051860. Review. ISSN: 1605-4806
2019	A Multi-Layered Study on Harmonic Oscillations in Mammalian Genomics and Proteomics.	Int J Mol Sci. 2019 Sep 17;20(18). pii: E4585. doi: 10.3390/ijms20184585.
2019	Generation of induced pluripotent stem cell line CSSi008-A (4698) from a patient affected by advanced stage of Dentato-Rubral-Pallidoluysian atrophy (DRPLA).	Stem Cell Res. 2019 Oct;40:101551. doi: 10.1016/j.scr.2019.101551. PMID:

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		31493762.
2019	Production and characterization of human induced pluripotent stem cells (iPSC) CSSi007-A (4383) from Joubert Syndrome.	Stem Cell Res. 2019 Jul;38:101480. doi: 10.1016/j.scr.2019.101480. PubMed PMID: 31202121
2019	Parkin Mutation Affects Clock Gene-Dependent Energy Metabolism.	Int J Mol Sci. 2019 Jun 5;20(11). pii: E2772. doi: 10.3390/ijms20112772. PubMed PMID: 31195749
2019	Transplantation of clinical-grade human neural stem cells reduces neuroinflammation, prolongs survival and delays disease progression in the SOD1 rats.	Cell Death Dis. 2019 Apr 25;10(5):345. doi: 10.1038/s41419-019-1582-5. PubMed PMID: 31024007
2019	DP71 and SERCA2 alteration in human neurons of a Duchenne muscular dystrophy patient.	Stem Cell Res Ther. 2019 Jan 15;10(1):29. doi: 10.1186/s13287-018-1125-5. PubMed PMID: 30646960
2018	Copy number variations in healthy subjects. Case study: iPSC line CSSi005-A (3544) production from an individual with variation in 15q13.3 chromosome duplicating gene CHRNA7	Stem Cell Res. 2018 Oct;32:73-77. doi: 10.1016/j.scr.2018.09.002. PMID: 30218896.
2018	Establishment of stable iPS-derived human Neural Stem Cells lines suitable for cell therapies	Cell Death Dis. 2018 Sep 17;9(10):937. doi: 10.1038/s41419-018-0990-2. PMID: 30224709
2018	Generation of the induced pluripotent stem cell line CSSi006-A (3681) from a patient affected by advanced-stage Juvenile Onset Huntington's Disease	Stem Cell Res. 2018 May;29:174-178. doi: 10.1016/j.scr.2018.04.008. PMID: 29704769
2018	Reciprocal interactions of mitochondria and the neuroimmunoendocrine system in neurodegenerative disorders: an important role for melatonin regulation	Front Physiol. 2018 Mar 12;9:199
2018	PRODUCTION AND CHARACTERIZATION OF CSSi003 (2961) HUMAN INDUCED PLURIPOTENT STEM CELLS (iPSCs) CARRYING A NOVEL PUNTIFORM MUTATION IN RAI1 GENE, CAUSATIVE OF SMITH–MAGENIS SYNDROME	Stem Cell Res. 2018 Apr;28:153-156. doi: 10.1016/j.scr.2018.02.016. PMID: 29494847
2018	Generation of induced pluripotent stem cell line, CSSi004-A (2962), from a patient diagnosed with Huntington's Disease at the presymptomatic stage	Stem Cell Res. 2018 Apr;28:145-148. doi: 10.1016/j.scr.2018.02.014. PMID: 29486399
2018	Generation of induced pluripotent stem cell line, CSSi002-A (2851), from a patient with juvenile Huntington Disease	Stem Cell Res. 2018 Mar;27:86-89. doi: 10.1016/j.scr.2018.01.011. PMID: 29342448
2018	Production and characterization of human induced pluripotent stem cells (iPSCs) from Joubert Syndrome: CSSi001-A (2850)	Stem Cell Res. 2018 Mar;27:74-77. doi: 10.1016/j.scr.2018.01.012. PMID: 29334628
2017	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects.	Am J Hum Genet. 2017 Oct 5;101(4):552-563. doi: 10.1016/j.ajhg.2017.08.017. PMID: 28965847

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2017	Glucose transportation in the brain and its impairment in Huntington disease: one more shade of the energetic metabolism failure?	Amino Acids. 2017 Jul;49(7):1147-1157.
2017	Alpha-7 Nicotinic Receptors in Nervous System Disorders: From Function to Therapeutic Perspectives	Cent Nerv Syst Agents Med Chem. 2017;17(2):100-108. doi: 10.2174/1871524916666160729111446. PMID: 27488345
2015	Stem cells from human amniotic fluid exert immunoregulatory function via secreted indoleamine 2,3-dioxygenase1 (IDO1).	J Cell Mol Med. 2015 Jul;19(7):1593-605. doi: 10.1111/jcmm.12534. PMID: 25783564
2013	Detrimental Effect of Class-selective Histone Deacetylase Inhibitors during Tissue Regeneration following Hindlimb Ischemia	J Biol Chem. 2013 Aug 9;288(32):22915-29. doi: 10.1074/jbc.M113.484337. PMID: 23836913
2013	A nitric oxide-dependent cross-talk between class I and III histone deacetylases accelerates skin repair.	J Biol Chem. 2013 Apr 19;288(16):11004-12. doi: 10.1074/jbc.M112.441816. PMID: 23463510
2012	P300/CBP Associated Factor Regulates Nitroglycerin-Dependent Arterial Relaxation by N{varepsilon}-Lysine Acetylation of Contractile Proteins	Arterioscler Thromb Vasc Biol. 2012 Oct;32(10):2435-43. doi: 10.1161/ATVBAHA.112.254011.
2011	Points to Epigenetics in Vascular Development	Cardiovasc Res. 2011 Jun 1;90(3):447-56. doi: 10.1093/cvr/cvr056. PMID: 21345806
2011	N{varepsilon}-lysine acetylation determines dissociation from GAP junctions and lateralization of connexin 43 in normal and dystrophic heart.	Proc Natl Acad Sci U S A. 2011 Feb 15;108(7):2795-800. doi: 10.1073/pnas.1013124108.
2011	Smad-Interacting Protein-1 and MicroRNA 200 Family Define a Nitric Oxide-Dependent Molecular Circuitry Involved in Embryonic Stem Cell Mesendoderm Differentiation.	Arterioscler Thromb Vasc Biol. 2011 Apr;31(4):898-907. doi: 10.1161/ATVBAHA.110.214478.
2010	Histone Deacetylase Inhibitors: Keeping Momentum For Neuromuscular And Cardiovascular Diseases Treatment.	*Pharmacol Res. 2010 Jul;62(1):3-10. doi: 10.1016/j.phrs.2010.02.014. PMID: 20227503*
2010	The Histone Deacetylase Inhibitor Suberoylanilide Hydroxamic Acid Improves Ventricular Arrhythmias In Dystrophic Mice.	Cardiovasc Res. 2010 Jul 1;87(1):73-82. doi: 10.1093/cvr/cvq035. PMID: 20164117
2010	Nitric oxide determines mesodermic differentiation of mouse embryonic stem cells by activating class IIa histone deacetylases: potential therapeutic implications in a mouse model of hindlimb ischemia.	*Stem Cells. 2010 Mar 31;28(3):431-42. doi: 10.1002/stem.300.PMID: 20073046*
2009	Nitric Oxide Deficiency Determines Global Chromatin Changes in Duchenne Muscular Dystrophy.	*FASEB J. 2009 Jul;23(7):2131-41. doi: 10.1096/fj.08-115618. PMID: 19264835*

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2008	HDAC2 blokade by nitric oxide and hystone deacetylase inhibitors reveals a common target in duchenne muscular dystrophy treatment.	Proc Natl Acad Sci U S A. 2008 Dec 9;105(49):19183-7. doi: 10.1073/pnas.0805514105.
2008	Nitric oxide modulates chromatin folding in human endothelial cells via protein phosphatase 2A activation and class II histone deacetylases nuclear shuttling.	Circ Res. 2008 Jan 4;102(1):51-8. doi: 10.1161/CIRCRESAHA.107.157305 . PMID: 17975112
2006	Non coding RNA and brain.	BMC Neurosci. 2006 Oct 30;7 Suppl 1(Suppl 1):S5. doi: 10.1186/1471-2202-7-S1-S5. PMID: 17118159
2003	Molecular recognition in helix-loop-helix and helix-loop-helix-leucine zipper domains. Design of repertoires and selection of high affinity ligands for natural proteins.	J Biol Chem. 2003 Apr 4;278(14):12182-90. doi: 10.1074/jbc.M211991200. PMID: 12514181
2002	Differential expression and localization of calmodulin-dependent phosphodiesterase genes during ontogenesis of chick dorsal root ganglion.	J Neurochem. 2002 Mar;80(6):970-9. doi: 10.1046/j.0022-3042.2002.00786.x.PMID: 11953447
2001	Making decisions through Myc	FEBS Lett. 2001 Feb 16;490(3):153-62 doi: 10.1016/s0014-5793(01)02118-4. PMID: 11223030

NATIONAL AND INTERNATIONAL COMMUNICATIONS (SELECTION)

Date	Title of Talk / Presentation	Conference Name and/or Organization	Venue and/or City
29 October 2022	SMS: De la biopsie au modèle cellulaire	Assemblée Générale Association Smith-Magenis,	Paris
29 September 2022	SMS:from biopsy to cell model.	Third scienti8c meeting “Smith-Magenis Families”	Rome
8-11 November 2021	Ipsc-derived neural stem cells from joubert syndrome as a novel in vitro model to elucidate ciliopathy-associated molecular mechanisms.	Neuroscience 2021 Virtual Presentation	Chicago, IL, USA
19-23 October 2019	Smith-magenis syndrome: in vitro and in vivo evaluation of ips-derived human neural stem cells for disease modeling and therapeutic approaches	Neuroscience 2021	Chicago, IL, USA

Date	Title of Talk / Presentation	Conference Name and/or Organization	Venue and/or City
3-7 November 2018	Establishment of stable iPS-derived human neural stem cells lines suitable for cell therapies and neurological diseases modeling	Neuroscience 2018	San Diego, CA, USA
11-15 November 2017	Establishment of stable, expandable and safe iPSCs derived-human neural stem cell lines suitable for cell therapies and disease modeling.	Neuroscience 2017	Washington DC, USA
15-18 November 2017	PATIENT SPECIFIC CELL LINES AS MODEL FOR SMITH-MAGENIS SYNDROME	SIGU	Naple, Italy
12-15 June 2017	HUMAN INDUCED PLURIPOTENT STEM CELLS (iPSCs) FROM JOUBERT SYNDROME AS A NOVEL in vitro MODEL TO ELUCIDATE CILIOPATHY-ASSOCIATED MOLECULAR MECHANISMS.	63° Convegno Gruppo Embriologico Italiano	Rome, Italy
14-18 November 2009	SIP1/ZEB2 Targeting by the Nitric Oxide-dependent miR200 Family is Important for Early Mesodermic/Cardiovascular Commitment of Mouse Embryonic Stem Cells"	American Heart Association meeting	Orlando, LA
8-12 November 2008	Mir429 Regulates NO-dependent Differentiation Of Mouse Embryonic Stem Cells	American Heart Association meeting	New Orleans, LA
1999	Dimerizzazione e funzione dei fattori trascrizionali bHLH(Zip): uso di repertori molecolari su fago	I FISV Conference	Riva del Garda

RESEARCH EXPERIENCE

1996-1997: We studied the level and characteristics of 3'-5'-cyclic nucleotide phosphodiesterase (PDE) activity in chick dorsal root ganglion (DRG) extracts of 5-day posthatching chicken (P5) and E10 and E18 embryos.

1998-2003: We investigated the molecular rules underlying recognition specificity of Helix-loop-helix (HLH) and helix-loop- helix-leucine zipper (HLHZip) and isolated molecules interfering with cell proliferation and differentiation control, two molecular repertoires on lambda phage capsids were obtained by directed randomization of the binding surface in these two domains were assembled. We used phage display technology to obtain interactors of the P75 receptor.

2004-2007: We studied small non-coding RNAs isolated and characterized from primary rat hippocampal cultures during neuronal differentiation.

2007-2011: Having observed the beneficial effect of deacetylase inhibitors and NO donors in dystrophic muscles, suggesting an unanticipated molecular link among dystrophin, NO signaling, and the histone deacetylases (HDACs), we demonstrated a special contribution of HDAC2 in the pathogenesis of Duchenne muscular dystrophy, indicating that HDAC2 inhibition by NO- dependent S-nitrosylation is important for the therapeutic response to NO donors in MDX mice. The role of nitric oxide in determining mesodermic differentiation of mouse embryonic stem cells by activating class IIa histone deacetylases was studied: potential therapeutic implications in a mouse model of hindlimb ischemia were demonstrated. The role of Smad- interacting protein-1 and microRNA 200 family in nitric oxide- dependent molecular circuitry involved in embryonic stem cell mesendoderm differentiation. To explore the epigenetic basis of Duchenne cardiomyopathy, dissociation from GAP junctions and lateralization of connexin 43 in normal and dystrophic heart determined by Ne-lysine acetylation was demonstrated. To address the role of epigenetic enzymes in the process of arterial vasorelaxation and nitrate tolerance, in vitro and in vivo experiments were performed in the presence or absence of glyceryl trinitrate (GTN) or histone deacetylases/histone acetylases modulators, demonstrating that P300/CBP associated factor regulates nitroglycerin-dependent arterial relaxation by N(e)-lysine acetylation of contractile proteins. The detrimental effect of class-selective histone deacetylase inhibitors during tissue regeneration following hindlimb ischemia was demonstrated.

2012-to date: My activity was focused on the production and characterization of Human Induced Pluripotent Stem Cells (iPSCs) from patients affected by various genetic diseases under virus-free and feeder-free conditions, suitable for cGMP certification. In my lab, we optimized a protocol to differentiate neural stem cells from human IPSC, with characteristics like neural stem cells obtained from the subventricular zone of human fetal brains and already used in clinical trials on ALS and MS-affected patients. The published protocol is suitable for cGMP certification. Over the years, we published several papers on primary fibroblasts obtained from patients affected by genetic disease. The fibroblasts of patients affected by Joubert syndrome showed that homozygous missense variants in SUFU gene significantly reduced SUFU stability and its capacity to bind GLI3 and promote its cleavage into the repressor form GLI3R. This deregulation impaired SUFU-mediated repression of the SHH pathway, resulting in recessive developmental defects of the CNS and limbs which share features with both SHH-related disorders and ciliopathies, in mice models. We investigated, in fibroblasts from genetic PD patients carrying parkin mutations, the interplay between mitochondrial bioenergetics and the cell autonomous circadian clock. Using two different in vitro synchronization protocols, we demonstrated that normal fibroblasts displayed rhythmic oscillations of both mitochondrial respiration and glycolytic activity. Conversely, in fibroblasts obtained from PD patients, a severe damping of the bioenergetic oscillatory patterns was observed. We assessed the ability of the six drugs, selected by computational analyses to directly bind purified σ1R in vitro by Surface Plasmon Resonance, to improve the growth of fibroblasts obtained from Huntington's Disease patients, which is significantly impaired with respect to control cells. All six of the selected drugs proved able to directly bind purified σ1R in vitro and improve the growth of HD cells from both or one HD patient. These results support the validity of the drug repositioning procedure for the identification of new therapeutic tools against HD. We also characterized primary cells derived from four SMS patients, two carrying the SMS-del, two with *RAI1* point mutations, and four control subjects to investigate pathogenetic processes underlying SMS. By combining transcriptomic and lipidomic analyses, we showed altered expression of lipid and lysosomal genes, deregulation of lipid metabolism, accumulation of lipid droplets, and a block of autophagy flux. Treatment with N-acetyl-cysteine (NAC) reduced cell death and lipid accumulation, suggesting new potential therapeutic targets for patient treatment. We optimized cellular reprogramming protocol and started to publish all induced pluripotent stem cell lines (iPSCs) we produced. The next step was the study of neural cells obtained from iPSCs. We investigated the Dp71 and Dp71- associated proteins cellular localization and expression in human neurons obtained by differentiation of induced pluripotent stem cell line from a patient affected by Duchenne Muscular Dystrophy with cognitive impairment. Moreover, we investigated the therapeutic potential of clinical-grade human neural stem cells (hNSCs) that have been successfully used in a phase I clinical trial for ALS patients (NCT01640067). We demonstrated that the beneficial effects observed after stem cell transplantation arise from multiple events that counteract several aspects of the disease, a crucial feature for multifactorial diseases, such as ALS suggesting that the combination of therapeutic approaches that target different pathogenic mechanisms of the disorder, including pharmacology, molecular therapy, and cell transplantation, will increase the chances of a clinically successful therapy for ALS.

LABORATORY EXPERIENCE

Molecular biology techniques: cloning, PCR, plasmidic DNA preparation, Lambda and M13 DNA preparation, electroporation, DNA sequencing, phage display library construction, colony hybridation, RNA extraction, episomal extraction, real time PCR, array of microRNA and PCR array, Northern Blotting, purification of proteins fused to GST and histidine tail, Affymetrix chips.

Cellular biology techniques: Cell culture, dissections, primary neuronal cell cultures, transfection in mammalian cells, transformation of mammalian cells, adenoviral and lentiviral infections, nucleofection, immunoprecipitation, GST and histidine pull-down, western blotting, immunohistochemistry, reprogramming technology, picking, embryoid bodies formation, neuralization, optimization of iPS differentiation protocols.

Biochemical techniques: PDE assay, HPLC and FPLC EMSA assay, Elisa assay, biopanning, immunoscreening, enzymatic assay (HDAC and HAT assay).

OTHER ROLES

Peer reviewer activity	2017- to date: peer reviewer activity for the following journals: Scientific Reports, Stem Cell Research & Therapy, AIMS Cell and Tissue Engineering, Cells, AJMS, Antioxidants, Biomolecules etc Revision of a research proposal submitted to the executive government agency of National Science Center Poland (NCN) Narodowe Centrum Nauki - NCN; http://www.ncn.gov.pl) 2022- to date: Guest editor of the special issue: Neural Stem Cells: Focusing on Disease Modeling and Translational Application". 2022 – to date: Review Editor for Neurogenomics in Frontiers in Neuroscience and Frontiers in Genetics 2023-to date: Associate Editor for Neurogenomics in Frontiers in Neuroscience and Frontiers in Genetics
Teaching Activity	2006 - Lecture in PhD Course in Cellular and Molecular Biology, University of L'Aquila, entitled: Study of protein-protein interaction through "phage display". 2013 - to date: academic supervision of PhD students and undergraduated students from University of Rome "La Sapienza" 2016 - Developmental Biology course, University La Sapienza, lesson entitled: induced pluripotent stem cells and their application.

	2017 – Accademia Medica di Roma, lesson entitled: 10 anni di iPS: stato dell'arte e le sfide che ci attendono 2018 - PhD course in Life Sciences and Biotechnology, University of Insubria. Lecture entitled: Cellule staminali neurali umane come sistema modello per lo studio di malattie neurodegenerative 2019 – Scuola Specializzazione di Neurologia, University of Palermo. Lecture entitled: Neural Stem cells as cellular model to study rare genetic disease of central nervous system. 2019 - PhD course in Biochemical Research, University of Rome. Lecture entitled: Human neural stem cells as a model to study neurodegenerative and neurodevelopmental diseases. 2023: external reviewer of doctoral thesis at Faculty of Health Sciences, UNIVERSITY OF EASTERN FINLAND. 2023: participation in the jury for the PhD Program in Biomedical Sciences, University of Padua.
MEMBERSHIP OF SCIENTIFIC ACADEMIES	2007-2011: member of the America Heart Association 2017-2022: member of the Society for Neuroscience 2024: SIGU Member
Workshop Organization	2002 - Scientific Board, "Third Meeting of Neuroscience Group of Cooperation", Rome

FOREIGN LANGUAGES

- Good level of **written** and **spoken English**

La sottoscritta dichiara di essere a conoscenza delle sanzioni penali cui incorre in caso di dichiarazione mendace o contenente dati non più rispondenti a verità, come previsto dall'art. 76 del D.P.R. 28.12.2000, n. 445. Il sottoscritto dichiara di essere a conoscenza dell'art. 75 del D.P.R. 28.12.2000, n. 445 relativo alla decadenza dai benefici eventualmente conseguenti al provvedimento emanato qualora l'Amministrazione, a seguito di controllo, riscontri la non veridicità del contenuto della suddetta dichiarazione.

Il sottoscritto, ai sensi del Regolamento UE 2016/679 e del D,Lgs. 196/2003, come da ultimo modificato dal D.Lgs. 101/2018, dichiara di essere a conoscenza che i propri dati saranno trattati dall'Università per assolvere agli scopi istituzionali ed al principio di pertinenza.

Rome, 19 September 2024