CURRICULUM VITAE ET STUDIORUM

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ResearcherID:	**R-8113-2018**
Scopus ID:	**9745766400**

Education

2004, January:	**Ph.D. Degree in Genetics and Molecular Biology**, University of Pavia, Italy
2004, January:	**Post-lauream Degree in Advanced School of Integrated Formation (S.A.F.I.),** University School for Advanced Studies (I.U.S.S) of Pavia, Italy
2000, July:	**Master Degree in Biological Sciences**, University of Pavia, Italy

Post graduate education and training

04/2015-present:	**Tenured Scientist, Researcher III Level –** CNR - Institute of Genetic and Biomedical Research (IRGB), UOS of Milan, Italy
06/2013-03/2015:	**Non Tenured Scientist, Researcher** (ex art.23) **–** CNR - Institute of Genetic and Biomedical Research (IRGB), UOS of Milan, Italy
05/2012-05/2013:	**Senior Post-Doctoral Fellow –**Laboratory of "Genoma Umano", directed by Dr. Paolo Vezzoni, CNR of Milan, Italy
01/2012-04/2012:	**Fondazione Telethon Co.Co.Co –** Project GGP10116 - Principal Investigator, Dr.ssa Cristina Sobacchi, Istituto Clinico Humanitas, Rozzano, Italy
21/10/2011:	**Habilitation as CNR Scientist, Researcher III Level**. Bando 364.92, Scientific Area G1.1 "Medical Sciences", homogeneous grouping II, Code ref. MI100/4
28/01/2010:	**Habilitation as CNR Scientist, Researcher III Level**. Bando 364.27, Area Scientifica (XII) "Scienze Mediche", Posizione C, Cod. ref. MI100/2
2006-2011	**Post-Doctoral Fellow –** Laboratory of "Genoma Umano", directed by Dr. Paolo Vezzoni, CNR of Milan, Italy
2004-2006:	**Post-Doctoral Fellow** at the Department of Genetics and Microbiology, Laboratory of Cytogenetics, directed by Prof. E. Raimondi, University of Pavia, Italy

2001-2004	**Graduate student** for Advanced School of Integrated Formation (S.A.F.I), University School for Advanced Studies (I.U.S.S.) of Pavia
2000-2003:	**Ph.D. fellow** at the Department of Genetics and Microbiology, Laboratory of Cytogenetics, directed by Prof. E. Raimondi, University of Pavia

Awards and grants

2020-2022:	**Grant recipient** -Progetto Regione Lombardia, INTERSLA "Innovazione, nuovi modelli tecnologici e reti per curare la SLA". Role: Coordinator of IRGB-CNR Unit.
2018-2019:	**Grant recipient** -Progetto Regione Lombardia, AMANDA "Alterazioni metaboliche, stress cellulari e processi neurodegenerative". Role: Coordinator of IRGB-CNR Unit.
01/2004	**Annual award recipient** - Advanced School of Integrated Formation (S.A.F.I), University School for Advanced Studies (I.U.S.S.) of Pavia.
12/2002	**Annual award recipient** - Advanced School of Integrated Formation (S.A.F.I), University School for Advanced Studies (I.U.S.S.) of Pavia.
05/2001	**Grant recipient -**"Progetto Giovani Ricercatori" M.U.R.S.T. -Ministero dell'Istruzione, dell'Università e della Ricerca- (Ministry of Instruction, University and Research).
12/2001	**Annual award recipient** - Advanced School of Integrated Formation (S.A.F.I), University School for Advanced Studies (I.U.S.S.) of Pavia.
11/2000	**Award recipient-** S.I.G.U. -Società Italiana Genetica Umana- (Italian Society of Human Genetic) - III Congress S.I.G.U., Orvieto, Italy
09/2000	**Award recipient -** "University curriculum" by I.S.U. -Istituto per il diritto allo Studio Universitario, University of Pavia.

Fellowship

05/2012-05/2013:	**Senior Post-Doctoral Fellow-** CNR of Milan, Laboratory of "Genoma Umano", directed by Dr. Paolo Vezzoni.
01/2012-04/2012:	**Fondazione Telethon Co.Co.Co –** Project GGP10116 - Principal Investigator, Dr.ssa Cristina Sobacchi, Istituto Clinico Humanitas, Rozzano, Italy
09/2006-12/2011:	**Post-Doctoral Fellow –**CNR of Milan, Laboratory of "Genoma Umano", directed by Dr. Paolo Vezzoni.
02/2005:	**Fondazione Adriano Buzzati Traverso**. Renew for 1 year, stipend support.
02/2004:	**Fondazione Adriano Buzzati Traverso**. 1 year, stipend support.

Teaching activity

2014	**Teaching in a training course for young researchers.** MbMM-Metodologie di base per l'innovazione nella diagnosi e nella terapia di Malattie Multifattoriali" Name of the course: Biomedical sciences- Stem cell biology (Scienze biomediche – Biologia delle cellule staminali). Accordo quadro regione Lombardia – CNR (2013-2015)
2006-2007	**Teaching assistant in Undergraduate Courses**. Name of the course: Human Genetics (Genetica Umana). Corso di studio in scienze Biotecnologie, University of Pavia

2004-2005:	**Teaching assistant in Undergraduate Courses**. Name of the Courses: Human Genetics (Genetica Umana), Corso di studio in scienze Biotecnologie; Genetic I and Genetic laboratory I (Genetica I e laboratorio di Genetica I). Corso di studio Interfacoltà in Biotecnologie, University of Pavia
2003-2004:	**Teaching assistant in Undergraduate Courses**. Name of the Course: Genetic II and Genetic laboratory (Genetica II e laboratorio di Genetica). Corso di studio Interfacoltà in Biotecnologie, University of Pavia
2002-2003:	**Teaching assistant in Undergraduate Courses**. Name of the Course: Genetic I and Genetic laboratory I (Genetica I e laboratorio di Genetica I). Corso di studio Interfacoltà in Biotecnologie, University of Pavia
2003-2007:	**Teaching Assistant** in "Genetics" for the University of Pavia. Cultore della Materia (Bio/18).
1999-2007:	**Tutoring and Supervision** for undergraduate students in Biology and Biotechnology.

Research areas and scientific activity

She has a long lasting research experience and expertise in the field of classical and molecular cytogenetic of mammalian cells and years of experience in mouse and human pluripotent stem cells (both embryonic stem cells and iPSCs). Her main interest is in the investigation of gene therapy approaches for genome disorders due to chromosomal abnormalities. A recent important part of her research has been focused on the use of the CRISPR/Cas9 technology in ESCs to induce the correction of recessive genetic defects by the endogenous repair machinery. Currently, her main activity is the generation and corrections of iPSCs from patients affected by genetic diseases and in iPSC differentiation with particular interest in the generation of iPSC-derived human organoids to help bridge gap between laboratory study and animal models.

Additional Information

Communication: Excellent communication skills and predisposition to teach, developed through training Ph.D. students both in laboratory and during classes. Good skills in presenting subject during meeting or public presentation (e.g. lessons, seminars, meeting, etc...)

Digital Competence: Good informatics proficiency for common-use software and internet navigation (e.g. Office, internet browsers, e-mail software, etc...); Bio-informatics tools (e.g. NCBI databases, primer design software, sequence alignment, software programs for generating sgRNAs and finding their target)

Languages: Italian, native language; English, fluently (writing, reading and oral expression).

Publications

Author of 21 scientific publications in peer-reviewed journals, 1 book chapter, and more than 20 proceedings of conferences.

H-index = 12 (Google Scholar) with 460 citations; 11 Web of Science; 12 Scopus (Author ID: 9745766400).

1. Mazzara PG, Muggeo S, Luoni M, Massimino L, Zaghi M, Valverde PT, Brusco S, Marzi MJ, Palma C, Colasante G, Iannielli A, **Paulis M**, Cordiglieri C, Giannelli SG, Podini P, Gellera C, Taroni F, Nicassio F,

Rasponi M, Broccoli V. Frataxin gene editing rescues Friedreich's ataxia pathology in dorsal root ganglia organoid-derived sensory neurons. Nat Commun. 2020 Aug 21;11(1):4178. doi: 10.1038/s41467-020-17954-3. PMID: 32826895; PMCID: PMC7442818.

2. **Paulis M.**, Susani L, Castelli A, Suzuki T, Hara T, Straniero L, Duga S, Strina D, Mantero S, Caldana E, Sergi LS, Villa A, Vezzoni P. Chromosome Transplantation: A Possible Approach to Treat Human X-linked Disorders. Mol Ther Methods Clin Dev. 2020 Jan 21;17:369-377. doi: 10.1016/j.omtm.2020.01.003. eCollection 2020 Jun

3. Salvarani N, Crasto S, Miragoli M, Bertero A, **Paulis M**, Kunderfranco P, Serio S, Forni A, Lucarelli C, Dal Ferro M, Larcher V, Sinagra G, Vezzoni P, Murry CE, Faggian G, Condorelli G, Di Pasquale E. The K219T-Lamin mutation induces conduction defects through epigenetic inhibition of SCN5A in human cardiac laminopathy. Nat Commun. 2019 May 22;10(1):2267. doi: 10.1038/s41467-019-09929-w.

4. Castelli A, Susani L, Menale C, Muggeo S, Caldana E, Strina D, Cassani B, Recordati C, Scanziani E, Ficara F, Villa A, Vezzoni P, **Paulis M.** Chromosome Transplantation: Correction of the Chronic Granulomatous Disease Defect in Mouse Induced Pluripotent Stem Cells. Stem Cells. 2019 Jul;37(7):876-887. doi: 10.1002/stem.3006. Epub 2019 Apr 2.

5. Crociara P., Chieppa M.N., Costassa E.V., Berrone E., Gallo M., Faro M.L., Pintore M.D., Iulini B., D'Angelo A., Perona G., Botter A., Formicola D., Rainoldi A., **Paulis M.**, Vezzoni P., Meli F., Peverali F.A., Bendotti C., Trolese M.C., Pasetto L., Bonetto V., Lazzari G., Duchi R., Perota A., Lagutina I., Quadalti C., Gennero M.S., Dezzutto D., Desiato R., Boido M., Ghibaudi M., Valentini M.C., Caramelli M., Galli C., Casalone C., Corona C. Motor neuron degeneration, severe myopathy and TDP-43 increase in a transgenic pig model of SOD1-linked familiar ALS. Neurobiol Dis. 2018, S0969-9961(18)30752-6. doi: 10.1016/j.nbd.2018.11.021

6. Susani L., Castelli A., Lizier M., Lucchini F., Vezzoni P., **Paulis M.** Correction of a Recessive Genetic Defect by CRISPR-Cas9-Mediated Endogenous Repair. The CRISPR Journal; 2018, 1(3):230-238. PMID: 28191778

7. Meneghini V., Frati G., Sala D., De Cicco S., Luciani M., Cavazzin C., **Paulis M.**, Mentzen W., Morena F., Giannelli S., Sanvito F., Villa A., Bulfone A., Broccoli V., Martino S., Gritti A. Generation of Human Induced Pluripotent Stem Cell-Derived Bona Fide Neural Stem Cells for Ex Vivo Gene Therapy of Metachromatic Leukodystrophy. Stem Cells Translational Medicine; 2017, 6:352–368. doi.org/10.5966/sctm.2015-0414

8. Lizier M., Anselmo A., Mantero S., Ficara F., **Paulis M.**, Vezzoni P., Lucchini F., and Pacchiana G. Fusion between cancer cells and macrophages occurs in a murine model of spontaneous neu+ breast cancer without increasing its metastatic potential. Oncotarget; 2016, 7(38): 60793–60806. doi: 10.18632/oncotarget.11508

9. **Paulis M**., Castelli A., Susani L., Lizier M., Lagutina I., Focarelli M.L., Recordati C., Uva P., Faggioli F., Neri T., Scanziani E., Galli C., Lucchini F., Villa A., Vezzoni P. Chromosome transplantation as a novel approach for correcting complex genomic disorders. Oncotarget; 2015, 6(34):35218-30. doi: 10.18632/oncotarget.6143.

10. Neri T., Muggeo S., **Paulis M**., Caldana M.E., Crisafulli L., Strina D., Focarelli M.L., Faggioli F., Recordati C., Scaramuzza S., Scanziani E., Mantero S., Buracchi C., Sobacchi C., Lombardo A, Naldini L, Vezzoni P, Villa A, Ficara F. Targeted Gene Correction in Osteopetrotic-Induced Pluripotent Stem Cells for the Generation of Functional Osteoclasts. Stem Cell Reports; 2015, 5(4):558-68. doi:10.1016/j.stemcr.2015.08.005.

11. **Paulis M**., Castelli A., Lizier M., Susani L., Lucchini F., Villa A., Vezzoni P. A pre-screening FISH-based method to detect CRISPR/Cas9 off-targets in mouse embryonic stem cells. Sci Rep; 2015, 5:12327. doi: 10.1038/srep12327.

12. Santin G., **Paulis M**., Vezzoni P., Pacchiana G., Bottiroli G., Croce A.C. Autofluorescence properties of murine embryonic stem cells during spontaneous differentiation phases. Lasers Surg Med; 2013, 45(9):597-607. doi: 10.1002/lsm.22182.

13. Lo Iacono N., Blair H.C., Poliani P.L., Marrella V., Ficara F., Cassani B., Facchetti F., Fontana E., Guerrini M.M., Traggiai E., Schena F., **Paulis M**., Mantero S., Inforzato A., Valaperta S., Pangrazio A., Crisafulli L., Maina V., Kostenuik P., Vezzoni P., Villa A., Sobacchi C. Osteopetrosis rescue upon RANKL administration to Rankl(-/-) mice: a new therapy for human RANKL-dependent ARO. J Bone Miner Res; 2012, 27(12):2501-10. doi: 10.1002/jbmr.1712.

14. Marrella V., Poliani P.L., Fontana E., Casati A., Maina V., Cassani B., Ficara F., Cominelli M., Schena F., **Paulis M.**, Traggiai E., Vezzoni P., Grassi F., Villa A. Anti-CD3ε mAb improves thymic architecture and

prevents autoimmune manifestations in a mouse model of Omenn syndrome: therapeutic implications. Blood; 2012, 120(5):1005-14. doi: 10.1182/blood-2012-01-406827.

15. **Paulis M.** Chromosome transfer via cell fusion. Methods Mol Biol; 2011, 738:57-67.
16. Revenkova E, Focarelli ML, Susani L, **Paulis M.**, Bassi MT, Mannini L, Frattini A, Delia D, Krantz I, Vezzoni P, Jessberger R, Musio. Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. A. Hum Mol Genet; 2009, 18(3):418-27.
17. Focarelli ML, Soza S, Mannini L, **Paulis M.**, Montecucco A, Musio A. Claspin inhibition leads to fragile site expression. Genes Chromosomes Cancer; 2009, 48(12):1083-90.
18. **Paulis M**, Bensi M, Orioli D, Mondello C, Mazzini G, D'Incalci M, Falcioni C, Radaelli E, Erba E, Raimondi E, De Carli L. Transfer of a human chromosomal vector from a hamster cell line to a mouse embryonic stem cell line. Stem Cells; 2007, 25(10):2543-50.
19. Uboldi C., **Paulis M.,** Guidi E., Bertoni A., Meo G.P., Perucatti A., Iannuzzi L., Raimondi E., Brunner R.M., Eggen A., Ferretti L. Cloning of the bovine prion-like Shadoo (SPRN) gene by comparative analysis of the predicted genomic locus. Mamm Genome; 2006, 17(11):1130-9.
20. **Paulis M.,** Bensi M., Moralli D., De Carli L. and Raimondi E. A Set of Duplicons on Human Chromosome 9 is Involved in the Origin of a Supernumerary Marker Chromosome. Genomics; 2006, 87(6):747-757.
21. Uboldi C., Del Vecchio I., Foti M.G., Azzalin A., **Paulis M.**, Raimondi E., Vaccari G., Agrimi U., Di Guardo G., Comincini S., Ferretti L. Prion-like Doppel gene (PRND) in the goat: genomic structure, cDNA and polymorphisms. Mammalian Genome; 2005, 16: 963-971.
22. **Paulis M.**, Moralli D., Bensi M., De Carli L. and Raimondi E. Isolation from the horse genome of a new DNA transposon belonging to the *Tigger* family. Mammalian Genome; 2004, 15(5): 399-403.

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Marianna Paulis