BIOGRAPHICAL SKETCH

NAME	POSITION TITLE
Monfregola, Jlenia	Senor post doc

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	YEAR(s)	FIELD OF STUDY
University of Naples "Federico II", Italy	B. SC.	2001	Gene regulation - Genomics
University of Naples "Federico II", Italy	Ph.D.	2006	Cell Biology, Cell metabolism
IGB-CNR Adriano Buzzati Traverso institute	Post Doc	2016-2009	Cell Biology, Vesicle Trafficking, Human diseases
The Scripps Research Institute, La Jolla (CA)	Research associate	2010-2013	Vesicle Trafficking, Human diseases
TIGEM, Pozzuoli (NA), Italy	Research associate	2014-pres	Human diseases, Genome editing

A. PERSONAL STATEMENT

During my research I have been always interested in approaches of Functional genomics and Cell biology to study human diseases. During the first part of my career, I worked, as part of the Human Genome Project, on the identification and characterization of new human genes with unknow function. At that time, I identified and characterize two new genes: the *TMLH gene, which* codifies for the ε-N-Trimethyllysine Hydroxylase, the first enzyme of carnitine biosynthesis and the *WASH* gene, that codify for a new member of the Wiskott-Aldrich syndrome protein (WASP)/WAVE family. The members of this family are NPF (Nucleation Promoting Factors) assembly factor necessary for the production of new actin branched filaments from pre-existing actin filaments. Defects on these genes are causative of the immune Wiskott-Aldrich syndrome. To expand my knowledge in studying the molecular basis of the mechanisms regulating actin remodeling and vesicular trafficking and their relationship to human disease, I moved to USA and I joined the laboratory of Dr. Sergio Catz' lab at Scripps, La Jolla. During my time at Dr. Catz's lab I have been working on different cellular and mouse models of immune disease and acquired some of the many skills required to study this fascinating field of biological research. A that point of my career, I decided to follow my major interest in functional genomics I joined the TIGEM institute as senor post doc in the laboratory of Prof. Andrea Ballabio. Thanks to my expertise in cellular biology and genomics I have been involved in the ambitious project to generate a biobank of cellular model to perform the first comparative study of a class of the human inerited disorder: the Lysosomal storage diseases. Within this project, I have built my expertise in the CRISPr technology and its application in vitro and in vivo.

B. POSITIONS AND HONORS:

Positions and Employment

2000-2001 Undergraduate, Institute of Genetics and Biophysics, CNR, Naples, Italy
2002-2006 PhD Student, Institute of Genetics and Biophysics, CNR, Naples, Italy
2006-2009 Postdoc. Fellow, Institute of Genetics and Biophysics, CNR, Naples, Italy
2010-2013 Res. associate, The Scripps Research Institute, La Jolla (CA)
2014-Pres Res. associate, Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli, Italy

Honors and Fellowships

2001 Magna cum laude, University of Naples Federico II, Italy.
2005 Institute of Genetics and Biophysics fellowship for the PhD program, Naples, Italy.
2006 Short EMBO fellowship University of Helsinki, Helsinki (Finland)
2008 Banco Napoli fellowship.
2011-2013 American Heart Association fellowship, TSRI (CA)
2014-2015 DTI-IMPORT/Marie Curie COFUND postdoctoral fellowship, TIGEM (IT)

PUBLICATIONS:

1. Ambrosio R, Fimiani G, **Monfregola J**, Sanzari E, De Felice N, Salerno MC, Pignata C, D'Urso M, Ursini MV.
The structure of human STAT5A and B genes reveals two regions of nearly identical sequence and an alternative tissue specific STAT5B promoter.
Gene. 2002 Feb 20;285(1-2):311-8.

2. Crispi S, Sanzari E, **Monfregola J**, De Felice N, Fimiani G, Ambrosio R, D'Urso M, Ursini MV.
Characterization of the human STAT5A and STAT5B promoters: evidence of a positive and negative mechanism of transcriptional regulation.
FEBS Lett. 2004. Mar 26;562(1-3):27-34.

3. Santoro A, Lioi MB, **Monfregola J**, Salzano S, Barbieri R, Ursini MV
L-Carnitine protects mammalian cells from chromosome aberrations but not from inhibition of cell proliferation induced by hydrogen peroxide.
Mutat Res. 2005 Nov 10; 587(1-2):16-25. 2005 Sep 15.

4. Monfregola J, Cevenini A, Terracciano A, van Vlies N, Arbucci S, Wanders RJ, D'Urso M, Vaz FM, Ursini MV.
Functional analysis of TMLH variants and definition of domains required for catalytic activity and mitochondrial targeting.
J Cell Physiol. 2005 . Sep;204(3):839-47

5. Monfregola J, Napolitano G, Conte I, Cevenini A, Migliaccio C, D'Urso M, Ursini MV. *Functional characterization of the TMLH gene: promoter analysis, in situ hybridization, identification and mapping of alternative splicing variants.*
Gene . 2007 Jun 15;395(1-2):86-97.

6. Laperuta C, Spizzichino L, D'Adamo P, **Monfregola J**, Maiorino A, D'Eustacchio A, Ventruto V, Neri G, D'Urso M, Chiurazzi P, Ursini MV, Miano MG.
MRX87 family with Aristaless X dup24bp mutation and implication for polyAlanine expansions.
BMC Med Genet . 2007 May 4;8:25.

7. Napolitano G, Mirra S, **Monfregola J**, Lavorgna A, Leonardi A, Ursini MV.

NESCA: A new NEMO/IKKgamma and TRAF6 interacting protein.
J Cell Physiol. 2009 Aug;220(2):410-7.

8. Monfregola J, Napolitano G, D'Urso M, Lappalainen P, Ursini MV.
Functional characterization of Wiskott Aldrich Syndrome protein and scar homolog (WASH), a bi-modular nucleation promoting factor (NPF) able to interact with biogenesis of lysosome related organelle subunit 2 (BLOS-2) and {gamma}-tubulin.
J Biol Chem. 2010 May 28;285(22):16951-7

9. Johnson JL, Hong H, **Monfregola J**, Kiosses WB, Catz SD. *MUNC13-4 restricts motility of RAB27A-expressing vesicles to facilitate lipopolysaccharide-induced priming of exocytosis in neutrophils.*
J Biol Chem. 2010 Dec 9.

10. Johnson JL, Hong H, **Monfregola J**, Catz SD. *Increased survival and reduced neutrophil infiltration of the liver in Rab27a- but not Munc13-4-deficient mice in lipopolysaccharide-induced systemic inflammation.* Infect Immun. 2011 Sep;79(9):3607-18.

11. Johnson JL, **Monfregola J,** Napolitano G, Kiosses WB, Catz SD. *Vesicular trafficking through cortical actin during exocytosis is regulated by the Rab27a effector JFC1/Slp1 and the RhoA-GTPase-activating protein Gem-interacting protein*. Mol Biol Cell. 2012 May;23(10):1902-16.

12**. Monfregola J,** Johnson JL, Meijler MM, Napolitano G, Catz SD**.**
Munc13-4 regulates the oxidative response and is essential for phagosomal maturation and bacterial killing in neutrophils. J Biol Chem. 2012 Oct 31.

13. Napolitano G, Johnson JL, He J, Rocca CJ, **Monfregola J**, Pestonjamasp K, Cherqui S, Catz SD. *Impairment of chaperone-mediated autophagy leads to selective lysosomal degradation defects in the lysosomal storage disease cystinosis*. EMBO Mol Med. 2015 Feb;7(2):158-74.

14. He J, Johnson JL, **Monfregola J,** Ramadass M, Pestonjamasp K, Napolitano G, Zhang J, Catz SD. *Munc13-4 interacts with syntaxin 7 and regulates late endosomal maturation, endosomal signaling, and TLR9-initiated cellular responses.* Mol Biol Cell. 2016 Feb 1;27(3):572-87.

15. Di Malta C, Siciliano D, Calcagni A, **Monfregola J**, Punzi S, Pastore N, Eastes AN, Davis O, De Cegli R, Zampelli A, Di Giovannantonio LG, Nusco E, Platt N, Guida A, Ogmundsdottir MH, Lanfrancone L, Perera RM, Zoncu R, Pelicci PG, Settembre C, Ballabio A. *Transcriptional activation of RagD GTPase controls mTORC1 and promotes cancer growth.* Science. 2017 Jun 16;356(6343):1188-1192.

16. Bartolomeo R, Cinque L, De Leonibus C, Forrester A, Salzano AC, **Monfregola J**, De Gennaro E, Nusco E, Azario I, Lanzara C, Serafini M, Levine B, Ballabio A, Settembre C. *mTORC1 hyperactivation arrests bone growth in lysosomal storage disorders by suppressing autophagy.* J Clin Invest. 2017 Oct 2;127(10):3717-3729.

17. Napolitano G, Esposito A, Choi H, Matarese M, Benedetti V, Di Malta C, **Monfregola J**, Medina DL, Lippincott-Schwartz J, Ballabio A. *mTOR-dependent phosphorylation controls TFEB nuclear export*. Nat Commun. 2018 Aug 17;9(1):3312.

18. Napolitano G, Di Malta C, Esposito A, de Araujo MEG, Pece S, Bertalot G, Matarese M, Benedetti V, Zampelli A, Stasyk T, Siciliano D, Venuta A, Cesana M, Vilardo C, Nusco E, **Monfregola J**, Calcagnì A, Di Fiore PP, Huber LA, Ballabio A. *A substrate-specific mTORC1 pathway underlies Birt-Hogg-Dubé syndrome*. Nature. 2020 Sep;585(7826):597-602.

19. Soldati C, Lopez-Fabuel I, Wanderlingh LG, Garcia-Macia M, **Monfregola J**, Esposito A, Napolitano G, Guevara-Ferrer M, Scotto Rosato A, Krogsaeter EK, Paquet D, Grimm CM, Montefusco S, Braulke T, Storch S, Mole SE, De Matteis MA, Ballabio A, Sampaio JL, McKay T, Johannes L, Bolaños JP, Medina DL. Repurposing of tamoxifen ameliorates CLN3 and CLN7 disease phenotype. EMBO Mol Med. 2021 Oct 7;13(10):e13742. doi: 10.15252/emmm.202013742. Epub 2021 Aug 19

20. De Risi M, Tufano M, Alvino FG, Ferraro MG, Torromino G, Gigante Y, **Monfregola J**, Marrocco E, Pulcrano S, Tunisi L, Lubrano C, Papy-Garcia D, Tuchman Y, Salleo A, Santoro F, Bellenchi GC, Cristino L, Ballabio A, Fraldi A, De Leonibus E. Altered heparan sulfate metabolism during development triggers dopamine-dependent autistic-behaviours in models of lysosomal storage disorders. Nat Commun. 2021 Jun 9;12(1):3495