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Decreto Rettore Università di Roma "La Sapienza" n 4065/2019 del 19/12/2019

GRAZIA DANIELA RAFFA Curriculum Vitae (ai fini della pubblicazione)

Roma, 02/01/2020

Part II – Education

Туре	Year	Institution	Notes (Degree, Experience,)
University graduation	1999	Sapienza, University of Rome	Laurea (bachelor degree) in
			Biological Sciences
			110/110 cum laude
			Research under direction of
			Prof. Maurizio Gatti (Genetic
			analysis of spermatogenesis in
			Drosophila melanogaster)
Pre-doctorate training	1999	Sapienza, University of Rome	Fellowship "Luca Barone" by
			Accademia Nazionale Dei
			Lincei
PhD	1999-	Sapienza, University of Rome;	PhD in Genetics and Molecular
	2002	Cornell University, USA	Biology; Research under
		<u>-</u>	direction of Prof. Maurizio Gatti
			(Genetic and molecular analysis
			of telomere capping in
			Drosophila melanogaster). In
			this period I spent 12 months in
			the laboratory of Prof. Michael
			L. Goldberg at Cornell
			University

Part III – Appointments

IIIA – Academic Appointments

Start	End	Institution	Position
2010	То	Sapienza, University of Rome	Researcher BIO/18-Assistant
	date	Department of Biology and	Professor of Genetics
		Biotechnology	

IIIB - Research Appointments

Start	End	Institution	Position
2003	2004	Sapienza, University of Rome	Postdoctoral associate in the
			laboratory of Prof. Maurizio Gatti
2004	2006	The Scripps Research Institute La Jolla, CA, USA	Research associate in the laboratory of Prof. Michael N. Boddy (Control of genome stability and DNA damage

			checkpoint in <i>Schizosaccharomyces pombe</i>).
2006	2010	Sapienza, University of Rome	Postdoctoral associate in the laboratory of Prof. Maurizio Gatti (<i>Drosophila</i> telomere biology) supported by Fellowships sponsored by:
			Fondazione "Adriano Buzzati Traverso" (2006/2007)
			Fondazione "Cenci Bolognetti" (2007/2009)
			Italian Association for Cancer Research (AIRC) (2009/2010)
2015	2017	Stanford University Medical Center- Division of Hematology, Stanford, CA, USA	Invited Assistant Professor to conduct research in collaboration with Prof. Steven Artandi (18 months). (Investigating the role of TGS1 in telomere homeostasis)
2016	2016	Columbia University–Department of Pathology and Cell Biology, New York, USA	Research in collaboration with Prof. Livio Pellizzoni (3 months) (Investigating the role of TGS1 in SMA)

Part IV – Teaching experience

Year	Institution	Lecture/Course
From	Sapienza, University of Rome	Course: Genetics for the Laurea Triennale in
2012		Scienze Naturali (6CFU)
to date		

Part V - Society memberberships, Awards and Honors

Year	Title
2010	AGI-Associazione Genetica Italiana (member)
2006	Winner of an award by Italian Society of Biophysics and Molecular Biology (SIBBM)
	for best paper published in 2005

Part VI - Funding Information [grants as PI-principal investigator or I-investigator]

Year	Title	Program	Grant value
2013-	A Drosophila Model for Spinal	Telethon GPP13147	€ 117.100
2015	Muscular Atrophy (SMA) (PI)		
2015-	A Drosophila model for Spinal	Istituto Pasteur- Fondazione	€ 60.000
2017	Muscular Atrophy (SMA):	Cenci Bolognetti-Programmi	·
	identification and characterization	di ricerca "Under 40" 2015-	
	of Smn interactors and phenotypic	2017	

	modifiers. (PI)		
2015-	Exploiting the Drosophila model	AIRC IG 2014 to Maurizio	€ 210.000
2017	system to investigate the function of	Gatti	
	human proteins involved in		
	telomere maintenance. (I)		
2018-	Exploiting the Drosophila model	AIRC IG 2017 to Maurizio	€ 220.000
to date	system to investigate the function of	Gatti	
	human proteins involved in		
_	telomere maintenance.(I)		_

Part VII – Research Activities

Keywords
Telomeres
Drosophila
Capping proteins
Spinal Muscular
Atrophy
SMN
TGS1

T7

Brief Description

My research activity is focused on two main areas of investigation: 1) The control of telomere protection in Drosophila melanogaster. My research on telomeres stems from my PhD work, in collaboration with Prof. Maurizio Gatti at Sapienza University. We have identified 4 novel proteins, necessary to prevent telomere fusion in Drosophila and defined the composition and function of the Terminin telomere capping complex (Cicconi et al., 2017; Raffa et al., 2011; Raffa et al., 2010; Raffa et al., 2009; Raffa et al., 2005; Cenci et al., 2003). In a collaboration with Dr. Stefano Cacchione at Sapienza University, we are currently characterizing a collection of terminin interactors, with the aim of identifying novel telomeric proteins, whose human homologs might play a conserved role at telomeres. We have recently found that one of the terminin interactors, the TGS1 hypermethylase, is a negative regulator of telomerase activity and telomere length in human cells, thanks to an ongoing collaboration with Prof. Steven Artandi at Stanford University (Chen et al, in press).

2) Understanding the molecular mechanisms of Spinal Muscular Atrophy (SMA) pathogenesis through the identification of novel phenotypic SMN modifiers.

We recently developed a *Drosophila* model to identify novel genetic interactors of SMN (the causative factor for spinal muscular atrophy) and found that WDR79 ameliorates SMN loss of function phenotypes both in flies and worms, thanks to a collaboration with Dr. Elia Di Schiavi (CNR, Naples) (Di Giorgio et al., 2017). We are currently exploring the functional relationships between SMN and TGS1 in different models for SMA. We performed transcriptome studies in CRISPR-derived human cells deficient for TGS1, which led to the finding that TGS1 plays a novel role in the biogenesis of small nuclear RNAs. By combining studies in *Drosophila* and in human cells, our aim is to explore the functions SMN, TGS1 and their interactors, to elucidate how perturbations in the transcriptome induce neurological phenotypes relevant for disease.

I am author of 21 scientific papers, 7 as first author, 3 as last and/or corresponding author.

I reviewed manuscripts for Communications Biology and Nucleic Acids Research

I have been invited at national and international meetings, as speaker or coorganizer:

2017: Cold Spring Harbor Laboratory Meeting: Telomeres & Telomerase

(New York, USA). The role of TGS1 in the regulation of hTR biogenesis
(Invited speaker)
2018: Scuola di Genetica in Cortona: Citogenetica Molecolare e
Citogenomica. Quando i cromosomi vanno in crisi: citogenetica molecolare
della disfunzione telomerica (Invited speaker)
2019: JOINT MEETING AGI - SIMAG" (Cortona). Co-organizer of the
mini symposium "Genome Instability"

2020: EMBO workshop: Telomere function and evolution in health and disease (Portugal). Co-organizer

Part VIII – Summary of Scientific Achievements

Product type	Number	Data Base	Start	End
Papers [international]	20	Scopus	1999	2019
Papers [international]	1	Accepted paper, in press [§] , not yet in	2019	2019
		Scopus		

[§]This accepted paper is included in the list of the publications selected for evaluation (publication number 1), but it was not considered for the calculation of the <u>Total Impact Factor</u>.

Total Impact factor	156.3
Total Citations	702
Average Citations per Product	35.1
Hirsch (H) index	12
Normalized H index*	0.6
Average Impact Factor per Product	7.8
Impact Factor as first/co-first/last author or corresponding author (total 10 publications)	75

*H index divided by the academic seniority (20 years, since Laurea in Scienze Biologiche).

Part IX- Selected Publications

List of the publications selected for the evaluation. For each publication report title, authors, reference data, journal IF (if applicable), citations, press/media release (if any).

- Chen L, Roake CM, Galati A, Bavasso F, Micheli M, Saggio I, Schoeftner S, Cacchione S., Gatti M, Artandi SE and Raffa GD*. Loss of human TGS1 hypermethylase promotes increased telomerase RNA and telomere elongation Cell Reports, 2019. Paper accepted, in press (Letter of acceptance included in the PDF). IF 2018: 7.8; Number of citations: 0
 Roake CM, Chen L, Chakravarthy AL, Ferrell JE, Raffa GD and Artandi SE. Disruption of Telomerase RNA Maturation Kinetics Precipitates Disease.
 - Molecular Cell, 2019. 74(4): 688-700.e3.
 - IF 2018: 14.5; Number of citations: 3
- **3.** Di Giorgio ML, Esposito A, Maccallini P, Micheli E, Bavasso F, Gallotta I, Vernì F, Feiguin F, Cacchione S, McCabe BD, Di Schiavi E and **Raffa GD***. *WDR79/TCAB1 plays a conserved role in the control of locomotion and ameliorates phenotypic defects in SMA models.*

Neurobiology of Disease, 2017. 105: 42-50. **IF 2017: 5.2; Number of citations: 5**

- 4. Cicconi A, Micheli E, Vernì F, Jackson A, Gradilla AC, Cipressa F, Raimondo D, Bosso G, Wakefield JG, Ciapponi L, Cenci G, Gatti M, Cacchione S and Raffa GD*. The Drosophila telomere-capping protein verrocchio binds single-stranded DNA and protects telomeres from DNA damage response. Nucleic Acids Research, 2017. 45(6): 3068-3085.
 - IF 2017: 11.6 Number of citations: 10
- 5. Cipressa F, Morciano P, Bosso G, Mannini L, Galati A, Raffa GD, Cacchione S, Musio A and Cenci G. *A role for Separase in telomere protection*. Nature Communications, 2016. 7.
 IF 2016: 12.1 Number of citations: 12
- Cenci G, Ciapponi L, Marzullo M, Raffa GD, Morciano P, Raimondo D, Burla R, Saggio I and Gatti M. *The Analysis of Pendolino (peo) Mutants Reveals Differences in the Fusigenic Potential among Drosophila Telomeres.* PLoS Genetics, 2015. 11(6).

IF 2015: 6.7 Number of citations: 10

 Burla R, Carcuro M, Raffa GD, Galati A, Raimondo D, Rizzo A, La Torre M, Micheli E, Ciapponi L, Cenci G, Cundari E, Musio A, Biroccio A, Cacchione S, Gatti M and Saggio I. AKTIP/Ft1, a New Shelterin-Interacting Factor Required for Telomere Maintenance. PLoS Genetics, 2015. 11(6).

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IF 2015: 6.7 Number of citations: 17
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- Sechi S, Colotti G, Belloni G, Mattei V, Frappaolo A, Raffa GD, Fuller MT and Giansanti MG. GOLPH3 Is Essential for Contractile Ring Formation and Rab11 Localization to the Cleavage Site during Cytokinesis in Drosophila melanogaster. PLoS Genetics, 2014. 10(5).
 - IF 2014: 7.5 Number of citations: 28
- **9.** Raffa GD, Raimondo D, Sorino C, Cugusi S, Cenci G, Cacchione S, Gatti M and Ciapponi L. *Verrocchio, a Drosophila OB fold-containing protein, is a component of the terminin telomere-capping complex.*

Genes and Development, 2010. 24(15): 1596-1601.

IF 2010: 12.9 Number of citations: 45

Raffa GD, Siriaco G, Cugusi S, Ciapponi L, Cenci G, Wojcik E and Gatti M. *The Drosophila modigliani (moi) gene encodes a HOAP-interacting protein required for telomere protection*. Proceedings of the National Academy of Sciences of the United States of America, 2009. 106(7): 2271-2276.

IF 2009: 9.4 Number of citations: 37

- 11. Raffa GD, Wohlschlegel J, Yates Iii JR and Boddy MN. SUMO-binding motifs mediate the Rad60dependent response to replicative stress and self-association. Journal of Biological Chemistry, 2006. 281(38): 27973-27981. IF 2006: 5.8 Number of citations: 26
- 12. Raffa GD, Cenci G, Siriaco G, Goldberg ML and Gatti M. *The putative Drosophila transcription factor woc is required to prevent telomeric fusions*. Molecular Cell, 2005. 20(6): 821-831.
 IF 2005: 14.9 Number of citations: 48

* corresponding author Databases: Journal Citation Reports (Web of Science); Scopus

Signature Grazia Daniela Raffa

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