

## CURRICULUM VITAE

**Name:** Alessandra Rufini  
**Date of birth:** 17/01/1974  
**Nationality:** Italian  
**e-mail:** [rufini@med.uniroma2.it](mailto:rufini@med.uniroma2.it)

## PRESENT POSITION

2010- to date Senior investigator in the Laboratory of Signal Transduction, Department of Biomedicine and Prevention, Rare Disease Unit, University of Rome "Tor Vergata"  
<http://www.labst.org/>

## EDUCATION

1997 - 2001: Ph.D. in Immunology, University of Rome "La Sapienza", Italy.  
1992 - 1997: M.Sc. in Biology, University of Rome, "Tor Vergata" (110/110, with honors)

## PAST EXPERIENCES

2015-2022 Scientific Director at Fratagene Therapeutics srl  
<http://www.fratagene.com/>

2012-2018 University Contract Research, Department of Biomedicine and Prevention, University of Rome "Tor Vergata"  
Research topic: "New therapeutic approaches for Friedreich ataxia"

2005-2010 University Contract Research, Department of Experimental Medicine and Biochemical Sciences, University of Rome "Tor Vergata"  
Research topic: "Molecluar mechanisms of apoptosis"

## FELLOWSHIPS

2020-2021 Research Fellowship, University of Rome "Tor Vergata", Department of Biomedicine and Prevention. Research topic: "Etravirine mechanisms of action in Friedreich ataxia".

2010-2012 Post-doctoral Fellowship, University of Rome "Tor Vergata", Department of Experimental Medicine and Biochemical Sciences. Research topic: "New therapeutic strategies for Friedreich ataxia".

2009: Senior Fellowship from "Fondazione Santa Lucia" for the project: "Control of the expression and stability of frataxin, the protein defective in Friedreich's Ataxia".

2002-2004: Post-doctoral AIRC (Italian Association for Cancer Research) Fellowship for the project: "The role of non-receptor tyrosine kinases in Fas-induced apoptosis"

1998-99: EMBO short term Fellowship, at the EMBL (European Molecular Biology Laboratory), Heidelberg, Germany. Project title: "Identification of human cDNAs inhibiting or antagonizing caspase activity using a functional assay in fission yeast", under the supervision of Prof. Giulio Superti-Furga.

## TRAINING

2000: Training at Uppsala Biomedical Center, Sweden. Homology modeling studies under the supervision of Dr. Gerard Kleiweg.

1999: Selected to attend the EMBO (European Molecular Biology Organization) course on "Bioinformatics: from genome sequences to protein structures", held in Uppsala Biomedical Center, Sweden.

1997: Visiting graduate student at Brown University, Providence (RI), USA. Project title: "Amyloid beta secretion in neuronal cell lines", under the supervision of Prof. Cristina Alberini.

## SCIENTIFIC CONFERENCES

Invitation as speaker at international conferences, among which:

- 1st Annual Ubiquitin Research and Drug Discovery in Cancer Conference - Jan 27-28, 2011, San Diego, CA. Presentation title: "Preventing the UPS-dependent degradation of frataxin, the protein defective in Friedreich's Ataxia"
- International Ataxia Research Conference 2015, Windsor, England. Presentation title: "New therapeutic approaches to Friedreich Ataxia"
- Keystone Symposium on Ubiquitin Signaling, Jan 28 - Feb 1, 2018 Granlibakken Tahoe Tahoe City, California, USA. Presentation title: "The E3 Ligase RNF126 as a New Therapeutic Target for Friedreich Ataxia"
- International Ataxia Research Conference 2019, Washington, USA. Presentation title: "Etravirine enhances frataxin translation: a promising candidate for Friedreich ataxia therapy".

More than 20 poster presentations at international scientific congresses.

## SEMINARS

Invitation to present seminars at:

- Facultad de Farmacia y Bioquímica, Universidad de Buenos Aires, Argentina. October 24, 2014. Seminar title: "New Therapeutic Approaches to Friedreich Ataxia". Invited by Prof. Javier Santos.
- Division of Pharmaceutical, Pharmacy Administration and Social Sciences, Duquesne University School of Pharmacy and Graduate School of Pharmaceutical Sciences, Pittsburgh, PA, USA. January 27, 2022. Seminar title: "Therapeutic Approaches to Friedreich Ataxia". Invited by Prof. David Lapinsky.

## PATENTS

US Patent 10,442,779 "Compositions and methods for treating Friedreich's ataxia".

Inventors: Testi R., Incani O., Rufini A, De Martino G.

US Patent 10,426,775 "Methods for treating Friedreich's ataxia with etravirine".

Inventors: Rufini A., Alfedì G., Testi R.

## AWARDS

2017 Winner of the 2016-2017 BeHEARD Technology Prize from Rare Genomics for the development of a RNF126 RNAi mouse model by Charles River.

## GRANTS

2020-2021 Ataxia UK grant on the project: "Etravirine as a potential therapeutic for Friedreich ataxia".  
Role: Principal Investigator.

## TEACHING

2019-to date: Contract Professor of Immunology and Immunopathology in Medicine and Surgery Degree, Saint Camillus International University of Health and Medical Sciences  
SSD Med/04. Language: English.

2021-to date: Contract Professor of General Pathology in Dentistry and Dental Prosthetics Degree, Saint Camillus International University of Health and Medical Sciences  
SSD Med/04. Language: English.

2010-to date: Tutor for students and thesis supervision in: Cellular and Molecular Biology Degree, Evolution and human biology Degree, Medical Biotechnology Degree, University of Rome "Tor Vergata"; Human Nutrition Sciences Degree, Saint Camillus International University of Health and Medical Sciences.

Tutor for the PhD program in Immunology and Applied Biotechnology, University of Rome "Tor Vergata" (cycle XXIII, XXVII, XXVIII, XXX, XXXI, XXXIII)

## SCIENTIFIC REVISION

Reviewer for several international scientific journals with IF;  
Reviewer for the funding agency FARA (Friedreich Ataxia Research Alliance).

## LANGUAGES

Italian - Native language  
English - Full professional proficiency

## PUBLICATIONS

- Benvenuto, M., Angiolini, V., Focaccetti, C., Nardozi, D., Palumbo, C., Carrano, R., Rufini, A., Bei, R., Miele, M.T., Mancini, P., Barillari, G., Cirone, M., Ferretti, E., Tundo, G.R., Mutti, L., Masuelli, L., Bei, R. "Antitumoral effects of Bortezomib in malignant mesothelioma: evidence of mild endoplasmic reticulum stress in vitro and activation of T cell response in vivo".  
Biol Direct. 2023 Apr 17;18(1):17. doi: 10.1186/s13062-023-00374-w.
- Luffarelli, R., Panarello, L., Quatrana, A., Tiano, F., Fortuni, S., Rufini, A., Malisan, F., Testi, R., Condò, I. "Interferon Gamma Enhances Cytoprotective Pathways via Nrf2 and MnSOD Induction in Friedreich's Ataxia Cells"  
Int J Mol Sci. 2023 Aug 11;24(16):12687. doi: 10.3390/ijms241612687.
- Palumbo C, Benvenuto M, Focaccetti C, Albonici L, Cifaldi L, Rufini A, Nardozi D, Angiolini V, Bei A, Masuelli L and Bei R. "Recent findings on the impact of ErbB receptors status on prognosis and therapy of head and neck squamous cell carcinoma"  
Front Med (Lausanne). 2023 Feb 2;10:1066021. doi: 10.3389/fmed.2023.1066021..
- Rufini A, Malisan F, Condò I, Testi R.

“Drug Repositioning in Friedreich Ataxia”.

Front. Neurosci., 2022 Feb 9;16:814445. doi: 10.3389/fnins.2022.814445.

- Quatrana A, Morini E, Tiano F, Vancheri C, Panarello L, Romano S, Marcotulli C, Casali C, Mariotti C, Mongelli A, Fichera M, Rufini A, Condò I, Novelli G, Testi R, Amati F, Malisan F.  
“Hsa-miR223-3p circulating level is upregulated in Friedreich's ataxia and inversely associated with HCLS1 associated protein X-1, HAX-1”  
Hum Mol Genet. 2022 Jan 7; ddac005. doi: 10.1093/hmg/ddac005
- Vavla M, Arrigoni F, Toschi N, Peruzzo D, D'Angelo MG, Gandossini S, Russo A, Diella E, Tirelli S, Salati R, Rufini A, Condò I, Testi R, Martinuzzi A.  
“Sensitivity of Neuroimaging Indicators in Monitoring the Effects of Interferon Gamma Treatment in Friedreich'Ataxia”  
Front. Neurosci., 09 October 2020 | <https://doi.org/10.3389/fnins.2020.00872>
- Tiano F, Amati F, Cherubini F, Morini E, Vancheri C, Maletta S, Fortuni S, Serio D, Quatrana A, Luffarelli R, Benini M, Alfedì G, Panarello L, Rufini A, Toschi N, Frontali M, Romano S, Marcotulli C, Casali C, Gioiosa S, Mariotti C, Mongelli A, Fichera M, Condò I, Novelli G, Testi R, Malisan F.  
“Frataxin deficiency in Friedreich's ataxia is associated with reduced levels of HAX-1, a regulator of cardiomyocyte death and survival”.  
Hum Mol Genet. 2020 Jan 15. pii: ddz306. doi: 10.1093/hmg/ddz306
- Vavla M, D'Angelo MG, Arrigoni F, Toschi N, Peruzzo D, Gandossini S, Russo A, Diella E, Tirelli S, Salati R, Scarpazza P, Luffarelli R, Fortuni S, Rufini A, Condò I, Testi R, Martinuzzi A.  
“Safety and Efficacy of Interferon  $\gamma$  in Friedreich's Ataxia”.  
Mov Disord. 2020 Jan 13. doi: 10.1002/mds.27979.
- Alfedì G, Luffarelli R, Condò I, Pedini G, Mannucci L, Massaro DS, Benini M, Toschi N, Alaimo G, Panarello L, Pacini L, Fortuni S, Serio D, Malisan F, Testi R, Rufini A.  
“Drug repositioning identifies etravirine as a potential therapeutic for Friedreich ataxia”  
Mov Disord. 2019 Jan 9; 34:323-334.
- Castro IH, Ferrari A, Herrera MG, Noguera ME, Maso L, Benini M, Rufini A, Testi R, Costantini P, Santos J.  
“Biophysical characterisation of the recombinant human frataxin precursor”.  
FEBS Open Bio. 2018 Jan 25;8(3):390-405. eCollection 2018 Mar.
- Benini M, Fortuni S, Condò I, Alfedì G, Malisan F, Toschi N, Serio D, Massaro DS, Arcuri G, Testi R, Rufini A.  
“E3 Ligase RNF126 Directly Ubiquitinates Frataxin, Promoting Its Degradation: Identification of a Potential Therapeutic Target for Friedreich Ataxia”.  
Cell Rep. 2017 Feb 21;18(8):2007-2017.
- Cherubini F, Serio D, Guccini I, Fortuni S, Arcuri G, Condò I, Rufini A, Moiz S, Camerini S, Crescenzi M, Testi R, Malisan F.  
“Src inhibitors modulate frataxin protein levels”  
Human Molecular Genetics. 2015 Aug 1;24(15):4296-305.
- Rufini A, Cavallo F, Condò I, Fortuni S, De Martino G, Incani O, Di Venere A, Benini M, Massaro DS, Arcuri G, Serio D, Malisan F, Testi R.  
“Highly specific ubiquitin-competing molecules effectively promote frataxin accumulation and partially rescue the aconitase defect in Friedreich ataxia cells”.  
Neurobiol Dis. 2015 Mar;75:91-9.

- Guccini I, Serio D, Condò I, Rufini A, Tomassini B, Mangiola A, Maira G, Anile C, Fina D, Pallone F, Mongiardi MP, Levi A, Ventura N, Testi R, Malisan F.  
“Frataxin participates to the hypoxia-induced response in tumors”.  
Cell Death Dis. 2011 Feb 24;2(1):e123.
- Rufini A, Fortuni S, Arcuri G, Condò I, Serio D, Incani O, Malisan F, Ventura N, Testi R.  
“Preventing the ubiquitin-proteasome-dependent degradation of frataxin, the protein defective in Friedreich's ataxia”.  
Human Molecular Genetics. 2011 Apr 1;20(7):1253-61.
- Condò I, Malisan F, Guccini I, Serio D, Rufini A, Testi R.  
“Molecular control of the cytosolic aconitase/IRP1 switch by extramitochondrial frataxin”  
Human Molecular Genetics. 2010 Apr 1;19(7):1221-9.
- Condo' I, Ventura N, Malisan F, Rufini A, Tomassini B and Testi R.  
“In vivo maturation of human frataxin”.  
Human Molecular Genetics, 2007 Jul 1;16(13):1534-40
- Corsi S\*, Rufini A\*, Stagni V, Condo I, Matafora V, Bachi A, Bonifazi AP, Coppola L, Superti-Furga G, Testi R, Barila D.  
“Src kinase phosphorylates Caspase-8 on Tyr380: a novel mechanism of apoptosis suppression”.  
EMBO J. 2006 May 3;25(9):1895-905. \*: share first authorship
- Holcomb M, Rufini A, Barila D, Klemke RL.  
“Deregulation of proteasome function induces ABL-mediated cell death by uncoupling P130CAS and C-CRKL”.  
J Biol Chem. 2006 Feb 3;281(5):2430-40
- D. Barilà, A. Rufini, I. Condò, N. Ventura, K. Dorey, G. Superti-Furga and R. Testi.  
“Caspase-dependent cleavage of c-Abl contributes to apoptosis”  
Molecular and Cellular Biology. 2003 Apr;23(8):2790-9.
- Malisan F, Franchi L, Tomassini B, Ventura N, Condo I, Rippo MR, Rufini A, Liberati L, Nachtigall C, Kniep B, Testi R.  
“Acetylation suppresses the proapoptotic activity of GD3 ganglioside”.  
Journal of Experimental Medicine. 2002 Dec 16;196(12):1535-41.
- A. Rufini and R. Testi.  
“Lipid signaling in CD95-mediated apoptosis”.  
Kluwer Academic/Plenum Publisher. Subcellular Biochemistry. 2002; vol. 36 (Phospholipid metabolism in apoptosis):285-308. Review.
- M.R. Rippo, F. Malisan, L. Ravagnan, B. Tomassini, I. Condo', P. Costantini, S. A. Susin, A. Rufini, M. Todaro, G. Kroemer e R. Testi.  
“GD3 ganglioside directly targets mitochondria in a bcl-2-controlled fashion”.  
FASEB J 2000 Oct;14(13):2047-54
- M.R. Rippo, F. Malisan, L. Ravagnan, B. Tomassini, I. Condo', P. Costantini, S. A. Susin, A. Rufini, M. Todaro, G. Kroemer e R. Testi.  
“GD3 ganglioside as an intracellular mediator of apoptosis”  
European Cytokine Network 2000 Sep; 11(3): 487-8