



RESEARCH EXPERIENCES

- Dates
- name and address of supervisor
- Occupation or position held
- Main activity

October 2015 - present

Professor Clara Nervi, Università La Sapienza Roma, Dipartimento di Scienze e Biotechnologie Medico chirurgiche, corso della repubblica 69, Latina

PhD student

Identification and characterization of the role of Calreticulin protein and 3'untranslated region in normal and neoplastic hematopoiesis.

- Dates
- name and address of supervisor
- Occupation or position held
- Main activity

Mar 2014 - Aug 2014

Ph.D. Pekka Katajisto, Institute of Biotechnology, University of Helsinki, Viikinkaari 9, Helsinki, Finland

erasmus student for master thesis

Age-selective segregation of peroxisomes during stem cell division.

- Dates
- name and address of supervisor
- Occupation or position held
- Main activity

Jul 2013 - Feb 2014

Ph.D. Davide Corona, Dipartimento di Scienze e Tecnologie Molecolari e Biomolecolari, University of Palermo, viale delle Scienze, edificio 16, Palermo

apprentice

Study of the role of Ubcd1 in germline stem cells of drosophila.

- Dates
- name and address of supervisor
- Occupation or position held
- Main activity

Apr 2013 - Jul 2013

Ph.D. Agata Giallongo, Institute of Biomedicine and Molecular Immunology "Alberto Monroy", CNR, via Ugo La Malfa 153, Palermo

apprentice

Polysome isolation from MCF7 cells.

- Dates
- name and address of supervisor
- Occupation or position held
- Main activity

Mar 2012 - Oct 2012

Prof. Antonio Russo, AOUP "Paolo Giaccone", oncology department, University of Palermo, via del Vespro 129

apprentice for bachelor thesis

Analysis of JAK2 mutations in PV, TE and PMF patients

Education

- Dates
- Name and type of organisation providing education and training
- Title of qualification awarded

2012-2015

University of Palermo, Italy

Master degree in Medical Biotechnology and Molecular Medicine, Grade 110/110 cum laude and honourable mention

• Dates	2007-2012
• Name and type of organisation providing education and training	University of Palermo, Italy
• Title of qualification awarded	Bachelor degree in Biotechnology. Grade 105/110

Congress participations

• Dates	8-9/9/2016
• Location	Firenze, Italy
• Title of congress	Recent advances in understanding and treating chronic myeloid neoplasms
• Dates	20-23/9/2016
• Location	Rome, Italy
• Title of congress	FISV 2016 – XIV congress
• Dates	18/11/2016
• Location	Rome, Italy
• Title of congress	7th BeMM symposium
• Dates	6/6/2018
• Location	Palermo, Italy
• Title of congress	Droplet digital PCR scientific conference

Scientific activity

I became interested in the study of genetic abnormalities of hematologic diseases during my bachelor thesis internship, focusing on the research of JAK2 exon 12 mutations by Sanger Sequencing in those Myeloproliferative Neoplasm patients resulted negative for the JAK2 V617F mutation, assayed by realtime PCR. This research experience allowed me to learn the routine practice of genetic analysis, exploring the context of molecular diagnosis in Myeloproliferative Neoplasms and comparing the diagnostic power of different techniques to assay gene mutational status. Subsequently, I maintained my interest for genetics, addressing it to the regulation of gene expression and during my master degree course, I undertake an internship being part of a larger project focused on the study of the interaction miRNA:mRNA, in cooperation with Bioinformaticians, in order to realize a prediction algorithm of the interaction miRNA:mRNA, based on microarray expression data, combining multiple expression data. The part of the project on which I worked regarded the optimization of polysomal RNA isolation from MCF7 cell lines, to gain the best separation of polysomal RNA on sucrose gradient. In this internship I approached the issues of RNA isolation, aimed to study the interaction of miRNA:mRNA. Then I oriented my interest in the regulation mechanisms of molecular biology to the field of stem cells, carrying out a study on the epigenetic role of the enzyme Ubcd1 in ovary germline stem cells (GSCs) of model organism *Drosophila melanogaster*. Particularly I employed tissue specific-RNA interference to induce Ubcd1 knock down (KD) in ovaries and then to observe the effects on several GSCs stemness marker and the monoubiquitination of histone H2B (Ub-H2B). By evaluating morphological and epigenetic changes following Ubcd1 KD with immunofluorescence microscopy and stemness-associated histone modifications analysis, I demonstrated that Ubcd1 KD resulted in the impairment of GSCs ovary pool, together with the complete loss of Ub-H2B and the reduction of stemness epigenetic markers. My regard to stemness and differentiation resulted in my master degree thesis, a project conducted on a stem-like mammary cell line, to observe the age-dependent segregation of peroxisome during asymmetric stem cell division. By cloning peroxisomal tag that could be fluorescently labeled in vivo, and employing live imaging microscopy and FACS analysis, I observed that peroxisome are differently destined by stem cell to daughters according to their labelling-age, particularly keeping older peroxisome to the stem cell and transferring the young peroxisomes to the differentiating daughter. The experience matured in the field of molecular biology, epigenetics and stem cell allowed me to be admitted to the PhD program in Morphogenesis and Tissue Engineering, at the university of Rome - La Sapienza. During my PhD I carried out the study of 3'UTR role of Calreticulin (CALR) in normal and neoplastic hematopoiesis. By using unilineage culture of CD34+ driven to differentiate towards all myeloid lineages, we observed a

hematopoietic lineage-specific regulation of CALR. Moreover, amongst Myeloproliferative Neoplasms patients cohort we detected two non-canonical mutations of CALR in two patients with enhanced erythrocytosis and resembling to polycythemia vera, whereas canonical CALR mutations are routinely associated with the increase of platelets observed in essential thrombocythemia and primary myelofibrosis. Those CALR mutations affected 3'UTR and coding sequence and were predicted to alter 3'UTR folding structure. The CD34+ cell isolated from such patients showed an erythroid push, associating with JAK/STAT activation and CALR expression increase. We reproduced such findings even in *in vitro* model bearing such non-canonical CALR mutations generated by CRISPR-CAS, and suggesting a role of CALR 3'UTR in lineage-fate decision, with CALR 3'UTR disruption being associated to aberrant erythropoiesis *in vitro* and *in vivo*.

Publications

A novel germline mutation in the 3' untranslated region of Calreticulin gene induces JAK/STAT signaling activation and erythrocytosis

Quattrocchi A, Tomassini S, Billi M., Scerpa MC, Cenfra N, Gentile M, Maiorca C, Ceccherelli A, De Marinis E, Grignani F, Cimino G, Nervi C.

Haematologica. Volume: 103 Supplement: 3 Pages: S47-S48 2018

A novel germline CALR mutation affecting an evolutionary conserved region of 3'UTR in JAK2-negative siblings with Polycythemia vera

Quattrocchi A, Tomassini S, Scerpa MC, Cenfra N, Ceccherelli A, Pisanò S, De Marinis E, Gentile M, Maiorca C, Cimino G, Nervi C.

HemaSphere. 2(S1):1004-1005, June 2018.

HDAC inhibition by valproic acid decreases JAK2V617F levels in myeloproliferative neoplasms via up-regulation of MIR-101, *in vivo* and *in vitro*

De Marinis E, Cenfra N, Liberati D, Scerpa MC, Pagano F, Quattrocchi A, Tomassini S, Gentile M, Ceccherelli A, Cicalini A, Fantasia F, Cimino G, Nervi C

HemaSphere. 2(S1):255-256, June 2018.

Communications to congress

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|--------------------------|--|
| • Dates | 20-24/9/2017 |
| • Location | Bologna, Italy |
| • Title of congress | ABCD congress 2017 |
| • Title of communication | Characterization of CALR 3'UTR functions in normal hematopoiesis and myeloproliferative neoplasms (poster) |
| • Dates | 22-24/3/2018 |
| • Location | Salerno, Italy |
| • Title of congress | National PhD Meeting |
| • Title of communication | Characterization of CALR 3'UTR functions in normal hematopoiesis and myeloproliferative neoplasms (poster) |
| • Dates | 14-17/6/2018 |
| • Location | Stockholm, Sweden |
| • Title of congress | 23 rd Congress of EHA |
| • Title of communication | A novel germline CALR mutation affecting an evolutionary conserved region of 3'UTR in JAK2-negative siblings with Polycythemia Vera (abstract) |
| • Dates | 18-20/9/2018 |
| • Location | Rimini, Italy |
| • Title of congress | XV Congresso Nazionale della Società Italiana di Ematologia Sperimentale |
| • Title of communication | A novel germline mutation in the 3' Untranslated region of Calreticulin gene induces JAK/STAT signalling activation and erythrocytosis (Oral communication) |

Awards

**PERSONAL SKILLS AND
COMPETENCES**

SPECIALIZATION COURSE

Statistic course for researchers – GIMEMA foundation – 11-12/4/2016

MOTHER TONGUE

ITALIAN

OTHER LANGUAGES

ENGLISH

- | | |
|--------------------|----------------------|
| • reading skills | very good (B2, CEFR) |
| • writing skills | very good (B2, CEFR) |
| • verbal skills | very good (B2, CEFR) |
| • listening skills | very good (B2, CEFR) |

**TECHNICAL SKILLS AND
COMPETENCES**

Cell cultures.
Protein extracts.
Polysome isolation
Quantification of protein and nucleic acids
miRNA-mRNA interaction prediction tools
mRNA folding prediction and analysis
PCR and RT PCR.
Nested PCR
Plasmid modification with restriction enzymes
Cloning
Bacterial culture (E.coli)
Lentiviral packaging and infection
SNAP-tag technology
Western Blot.
Protein Immunoprecipitation.
Microscopy (fluorescence, confocal, structured illumination)

Classical genetic approaches.
Diagnostic kits.
DNA – RNA sanger sequencing analysis
Good knowledge of Microsoft Windows XP, Mac and Linux. Basis of R language.
Advanced use of GraphPad Prism.

Date 27.05.2019