NAME: Federica Carla Sangiuolo **POSITION TITLE**: Associate professor

PLACE AND DATE OF BIRTH: Naples, May 10th 1966

NATIONALITY: Italian

ACTUAL POSITION

Full Professor of Medical Genetics, Biomedicine and Prevention Department at "Tor Vergata" University of Rome.

Responsible of Molecular Genetics Unit at Tor Vergata Hospital, Department of Oncoematology

ADDRESS

Dipartimento di Biomedicina e Prevenzione Università di Roma Tor Vergata Via Montpellier 1 00133 ROMA(I) Tel. 06/72596164

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EDUCATION/TRAINING

Naples University (Italy) BS 1985-1988 Molecular Biology; Urbino University (Italy) BS 1988-1989 Molecular Genetics; Genoa University (Italy) Ph.D 1990-1994 Medical Genetics

POSITION AND EMPLOYMENT

1990 – 1995: Assistant Researcher of Human Genetics, Tor Vergata University of Rome;

1995 – 2001: Assistant Professor of Human Genetics, Tor Vergata University of Rome;

2001 – present: Associate Professor of Human Genetics, Tor Vergata University of Rome;

2009 - present: Director of Medical Genetics Postgraduate School, Tor Vergata University of

Rome

RESEARCH DOCTORATE

From 2009 to 2012:

member of ADVANCED TECHNOLOGIES IN BIOMEDECINE doctorate school at Tor Vergata University of Rome

From 2012 until today:

member of MEDICAL BIOTECHNOLOGY AND TRANSLATIONAL MEDECINE doctorate school at Tor Vergata University of Rome

MASTER SCHOOL

From 2011 she is Director of Master CYTOGENETICS AND CYTOGENOMICS From 2014 she teachs "Personalised Medecine: genetic test and nutrition" in Master: PERSONALISED NUTRITION: molecular and genetic basis.

▲ SCIENTIFIC ACTIVITY

Scientific activity is assessed by over 130 scientific papers on International journal with peer review.

Main research topics are:

- A Principal research issues regard the molecular study of genetic diseases such Cystic Fibrosis, beta-thalassemia, Thomsen/Becker Myotonia, Treacher Collins Syndrome, Galactosemia. At this regard molecular protocols of pre and post natal diagnosi were developed together with the characterization of new mutations or polymorphisms and population genetics study.
- ▲ Mapping of a gene causing a severe myopathy on chromosome 19
- ▲ Molecular characterisation of a new isoform of Lox gene, linked to myocardial infarction
- ▲ Optimisation of myocardial infarction viral and not viral gene therapy protocols focused to the correction of gene causing Cystic Fibrosis and Spinal Muscular Atrophy in different kind of cells

- ▲ Gene therapy protocols lentivirally mediated in embryonic stem cells derived from murine model of Spinal Muscular Atrophy
- ▲ Pharmacological treatments developed in vivo in murine models of Spinal Muscular Atrophy
- ▲ Culture and differentiation of human embryonic stem cells into type II pneumocytes; in vivo preclinical studies for pulmonary fibrosis
- ▲ Currently the research is focused on the creation and study of induced pluripotent cells (iPSCs) as a model of the pathogenesis of genetic diseases; further protocols have been optimized in order to identify residual undifferentiated cells by Volatile compounds analysis, aspect that results to be particularly useful in cell therapy approach.
- ▲ Evaluation of LOX-1 protein role for the insurgence and tumorigenesis of colon cancer, LOX-1 characterization as a novel biomarker and molecular target in therapeutic protocols for colon cancer.

GRANTS

1998: Principal Investigation of a Telethon Project: ISOLATION OF THE GENE CAUSING A MYOPATHY WITH RIMMED VACUOLES (MDVR), n.1076

2003: Principal Investigation of a Fondazione Fibrosi Cistica Project: Identification of CF modifier genes by family studies and microarray analysis

2003: Research Unit of a Project of Ministryof Research: Ricerca di modulatori genetici del fenotipo in Fibrosi Cistica

2003: Research Unit of a Project of Ministryof Research: Ricerca di modulatori genetici del fenotipo in Fibrosi Cistica

2004: Research Unit of a Fondazione Fibrosi Cistica Project: Screening of CFTR gene rearrangements in Italian CF patients

2005: Principal Investigator of a Fondazione Fibrosi Cistica Project: CFTR gene correction in human embryonic stem cells mediated by Small Fragment Homologous Replacement (SFHR)

2006: Principal Investigator of a FightSMA Foundation Project: SFHR-Mediated Modification of Murine ES-Derived SMA Motor Neurons: a Step Forward Through a Novel Cell-Therapy For Spinal Muscular Atrophy (SMA)

2009: Research Unit of a Ministry of Health Project: Caratterizzazione in vivo di marcatori d'espressione per la rivelazione del doping da IGF

2009: Research Unit of a Fondazione Roma Project: Molecular mechanisms in the pathogenesis of type 2 Diabetes mellitus and its cardiovascular complications

2009: Research Unit of a Fondazione Cenci Bolognetti Project: The interplay between epigenetics, cell cycle and homologous recombination in the gene therapy by Small Fragment Homologous Replacement (SFHR)

2011: project granted by European Project FP7-HEALTH-2009-single-stage: European multicenter network to evaluate pharmacokinetics, safety and efficacy of Meropenem in neonatal sepsis and meningitis

2014: Research Unit in a project granted by Italian Space agency entitled "COREA - COntromisure per le REAzioni degli astronauti"

2015: project granted Fondazione Roma and entitled" Sviluppo di un protocollo integrato su dati genetici/epigenetici ed ambientali per la predizione del rischio dell'infarto acuto del miocardio (AMI) in pazienti con aterosclerosi coronarica: studio pilota "

2016 : project granted by Foreign Affairs Ministry entitled "Undiagnosed diseases: a joint Italy-USA project"

2017 project granted by GFI (Grant for Fertility Innovation): "Endometrial Notch pathway as a novel target for improving implantation efficiency".

2018 project granted by NASA- MISSE-10 Polymers and Composites Experiment (PCE), champion M10N-C10.

2018 project granted by Regione Lazio: Sistema di misura dei composti volatili per la diagnosi invitro e in-vivo del carcinoma del colon (VOLACOL)

2019 project granted by Tor Vergata University: Diagnosi Precoce, Prognosi E Predittivita' Nel Cancro Del Colon-Retto: Lox-1 Come Target Nella Medicina Di Precisione (Propriocollox)