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DELLE MARCHE

**Multi-omics and AI approach in rare diseases:
implementing an innovative diagnostic pathway
and precision medicine tool for fibrotic diseases**

Prof. Gianluca Moroncini



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Supervisor: Prof. Gianluca Moroncini

Research Group Description: the Supervisor

Prof. Gianluca Moroncini, MD, PhD.

Gianluca Moroncini is **Full Professor of Internal Medicine** (SDS MED/09) at UNIVPM, and **Director of “Clinica Medica”** at Marche University Hospital, Ancona, Italy.

He is the Director of the Scleroderma Unit sponsored by the Italian Group against Scleroderma (GILS), the Head of EUSTAR (European Scleroderma Trials and Research) Centre 034, and the local representative of the European Reference Network (ERN) ReCONNET, focused on rare and low prevalence connective tissue and musculoskeletal diseases.

More than 90 publications on immuno-mediated diseases, especially systemic sclerosis.

<https://orcid.org/0000-0002-0380-0105>

Vice President of Marche Biobank and Director of the Marche Biobank Lab.

Grants

- 2022-2026: PNRR PE6 HEAL ITALIA – SPOKE 7 “Integrated and gender medicine approaches for prevention strategies based on environmental, lifestyle and clinical biometric data” (6.225M€). Principal Investigator.
- 2019-2023: POR FESR 2014-2020 “Marche BioBank – Collaborative research platform in the field of personalized medicine: drugs, diagnostics and new therapeutic approaches” (1.7 M€). Principal Investigator.
- 2018-present: Development of the optimal touchscreen interface for patients with scleroderma. The World Scleroderma Foundation and The French Association for Scleroderma first grant “Epidemiology & Quality of life in Systemic Sclerosis”. 25 K€. Principal investigator.
- 2016-2018: Systemic Sclerosis and Chronic Graft versus Host Disease: application of novel predictive assays and generation of transgenic animal models. Italian Ministry of Health. 371.000,00 €. Co-investigator.
- 2010-2011: GILS (Gruppo Italiano Lotta alla Sclerodermia). Validation of autoimmunity to PDGF Receptor as a pathogenic mechanism in scleroderma. 50 K€. Principal investigator.
- 2011: NIH, US, Grant 1R21AR055806-01A1 Experimental models of scleroderma pathogenesis. 250 K\$. Co-investigator.



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MARCHE BIOBANK

Marche BioBank, a 150 m² aptly designed space located at walking distance from Marche University Hospital, dedicated to the storage, characterization and distribution of samples and primary cell cultures and organoids obtained from patients with rare and chronic diseases, and with hematological-oncological malignancies.

STAFF: The Research Group is composed by one Full Professor (Prof Gianluca Moroncini), three Researchers (Dr. Devis Benfaremo, Dr. Matteo Mozzicafreddo, Dr. Silvia Agarbati), two Technicians (Dr. Silvia Svegliati Baroni, Dr. Chiara Paolini), two PhD students (Dr. Mario Piga, Dr. Carolina Clementi), one data manager (Dr. Norma Sartorelli)

Marche BioBank is part of the BBMRI-Biobanking and Biomolecular Resources Research Infrastructure (<https://www.bbmri.it/home>)



RESEARCH AND PUBLICATIONS

<https://orcid.org/0000-0002-0380-0105>

LABORATORY

Marche BioBank has a 150 m² associated laboratory endowed with single cell analysis facility (BD FACS Melody, Laser capture dissection microscope, digital PCR, Illumina NGS, Luminex, Mass spectrometer) for the complete omics characterization of liquid and tissue biopsies of patients.

Marche BioBank is part of a public-private partnership with the other Universities (UNIURB, UNICAM) and the 3 main biotech and pharmaceutical companies (Angelini, Diatheva, Diatech) of Marche Italian Region, which are actively contributing to ongoing research projects on precision medicine (www.marchebiobank.it)

**Dept. of Clinical and Molecular Sciences – DISCLIMO
UNIVPM**



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Supervisor: Prof. Gianluca Moroncini

**Department of Clinical and Molecular Sciences
(DISCLIMO)**





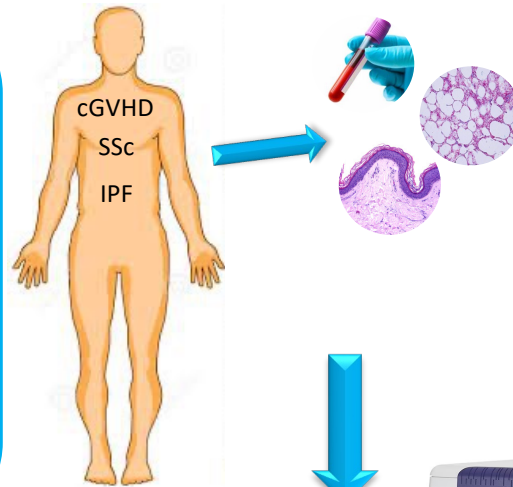
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Project Idea: Multi-omics and AI approach in rare diseases: implementing an innovative diagnostic pathway and precision medicine tool for fibrotic diseases

Background: Fibrosis is the late stage of many chronic, rare diseases characterized by immune-mediated inflammation such as Systemic Sclerosis (SSc), Idiopathic Pulmonary Fibrosis (IPF), Chronic Graft Versus Host Disease (cGVHD). Predicting fibrosis onset and progression is an unmet medical need. Resolving the heterogeneity of fibrotic disorders not only at early disease stages but also at later disease stages, through stratification of patients, could be achieved by multi-omics approach and artificial intelligence (AI) algorithms enabling integration of multi-level information (clinical, imaging, laboratory, omics) coming from a multitude of single patients.

Aim 1) Our network of centers dedicated to fibrotic diseases will provide liquid (blood, bronchoalveolar lavage) and tissue (skin, lung, heart, liver) biopsies, that will be transferred to Marche Biobank for extraction of single cell information both at DNA/RNA and protein level by multi-OMICS technologies.



Aim 2) Novel single cell data, conventional laboratory data, imaging data and clinical data from each patient will be integrated by bioinformatic/AI tools into new algorithm models enabling identification of new subsets of affected individuals across different diseases, stratified for risk of developing fibrosis, risk of progression to severe forms of fibrosis and possible response to new and existing targeted therapies.

