UNIVERSITÀ POLITECNICA DELLE MARCHE

Supervisor: Prof. Gianluca Moroncini

Dept. of Clinical and Molecular Sciences

Project idea: Biobanking and stratification of Rare Diseases

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Prof. Gianluca Moroncini, MD, PhD.

Gianluca Moroncini is **Full Professor of Internal Medicine** (SDS MED/09) **Director of Department of Clinical and Molecular Sciences**, UNIVPM and **Director of Clinica Medica,** Marche University Hospital, Ancona, Italy.

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

More than 100 publications on immune-mediated diseases, especially systemic sclerosis.

3 International patents

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

President of Marche Biobank SCARL and Director of Marche Biobank Lab.

Director of the Center for Precision Medicine on Rare Diseases – HEAL ITALIA

www.marchebiobank.it
https://officinemedicinadiprecisione.it/en/centers/centro-di-medicina-di-precisione-heal-italia-di-ancona/

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: 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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Research Group Description: the Group

MARCHE BIOBANK

Marche BioBank, located at walking distance from Marche University Hospital, is dedicated to the storage, characterization and distribution of samples and primary cell cultures and organoids obtained from patients with rare and chronic diseases.

***STAFF*:** The Research Group is composed by one Professor (Prof Gianluca Moroncini), three researchers (Dr. Devis Benfaremo, Dr. Silvia Agarbati,Dr. Matteo Mozzicafreddo), two technicians (Dr. Silvia Svegliati Baroni, Dr. Chiara Paolini), two PhD students (Dr. Carolina Clementi Dr. Barbara Perugini)

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

RESEARCH AND PUBBLICATIONS

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, Chromium Xo, Mass spectrometer) for the most complete omics characterization of liquid and tissue biopsies of patients.

Marche BioBank is a public-private partnership of 3 Universities (UNIVPM,UNIURB, UNICAM) and 3 biotech companies (Diatheva, Diatech, Mivell) in 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

UNIVERSITÀ POLITECNICA DELLE MARCHE

Supervisor: Prof. Gianluca Moroncini

Department of Clinical and Molecular Sciences (DISCLIMO)

SUN RISE

EXCELLENCE IN RESEARCH

MARCHEBIOBANK

16 SCIENTIFIC AREAS

BIO/17, MED/02, MED/04, MED/05, MED/06, MED/09, MED/12, MED/13, MED/15, MED/16, MED/31, MED/33, MED/35, MED/44, MED/46, MED/50

UNDERGRADUATES
Students of medicine and environmental and workplace prevention techniques
POSTGRADUATE MEDICAL EDUCATION
Allergology and Clinical Immunology; Clinical Pathology and Clinical Biochemistry; Dermatology and Venereology; Emergency Medicine; Endocrinology and Metabolic Diseases; Food Science; Geriatrics; Diseases of the digestive system; Haematology; Internal Medicine; Medical Oncology; Occupational Medicine; Orthopedics and Traumatology; Rheumatology.

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CLINICAL RESEARCH UNITS

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Project Idea: Biobanking and stratification of rare diseases

Background: Rare diseases, defined as those affecting fewer than 5 in 10,000 individuals, are in fact very numerous (over 6,000 distinct diseases, affecting a total of about 30 million people in Europe), extremely heterogeneous (very different endophenotypes are often grouped under the same designation), difficult to diagnose (diagnostic delays are much longer than for other diseases), and challenging to treat (they are often lacking specific therapies)

Objective 1. Biobanking

- Collect biological samples from patients with rare diseases
- Store samples in the Marche Biobank
- Enable multi-omics molecular analysis

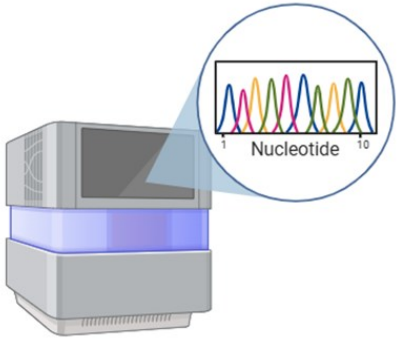
Objective 2. Data Integration & Stratification

- NGS transcriptomics
- Single-cell analysis
- Proteomics & immunophenotyping
- Tissue microdissection
- Clinical data integration
- AI-driven models for patient stratification

Digital PCR

Cell sorting

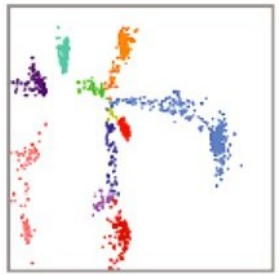
Single cell analysis

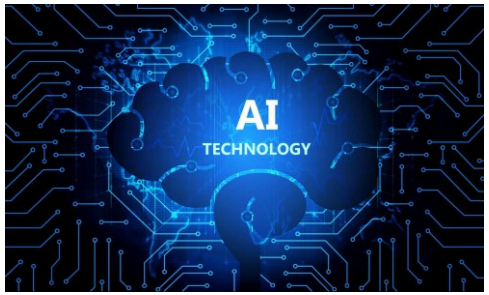


Next Generation Sequencing

Immunophenotyping

Tissue microdissection





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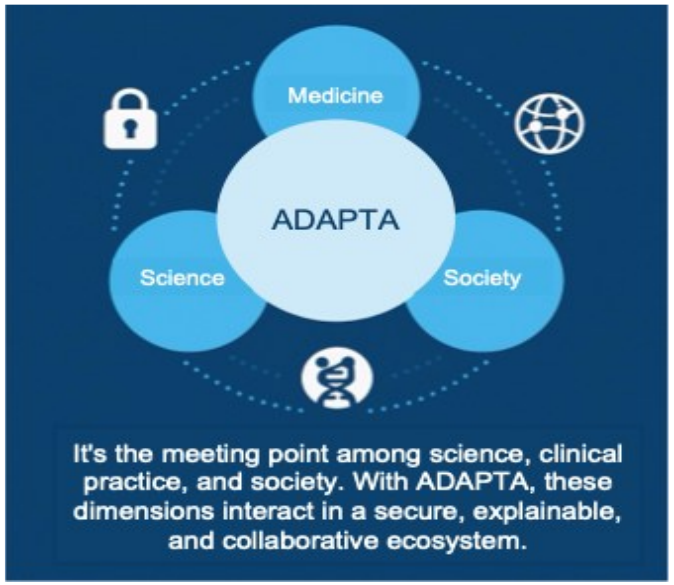
ADAPTA: A human-centric framework for data analysis in precision medicine

ADAPTA: AI-Driven Data Integration for Precision Medicine

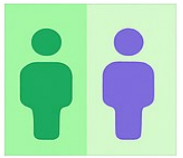
Novel single cell data, conventional laboratory data, imaging data and clinical data from each patient will be integrated into new algorithm models enabling identification of new subsets of affected individuals across different diseases, and possible response to new and existing targeted therapies.



Manage count tables + clinical data

Explore PCA plot + expression heatmap

Compare differential expression



Assess patient stratification

Expected outcomes:

- Integration of genetic, molecular, and clinical data
- Collaboration between clinicians, data scientists, and researchers
- Development of new tools for personalized treatment
- Improved patient care in rare diseases