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

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 (cGHVD). 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.

Research Group Description

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Networks: EUSTAR, ERN-RECONNET, ERN-LUNG, GITMO.
Facilities: Marche BioBank, 150 m2 aptly designed space located at walking distance between the UNIVPM Departments and Hospital, dedicated to the collection, characterization and distribution of samples and primary cell cultures and organoids obtained from patients with rare and chronic diseases, and with hematological-oncological malignancies. Marche BioBank is part of the BBMRI-Biobanking and Biomolecular Resources Research Infrastructure (https://www.bbmri.it/home).
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 most complete omics characterization of liquid and tissue biopsies of patients.
Public-private partnership between Marche BioBank and the 3 main biotech and pharmaceutical companies of Marche region (Angelini, Diatheva, Diatech) which are actively contributing to ongoing research projects on precision medicine.