

**To the Academic Board of the Research Doctorate in Molecular Medicine**

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*Scientific activity carried out in the 1<sup>st</sup> year of PhD, Academic Year 2020/2021*

**Introduction**

Multiple myeloma (MM) is a malignancy of differentiated plasma cells and it is still incurable. It is important to distinguish symptomatic MM from the precursor stages of monoclonal gammopathy of undetermined significance (MGUS) and smoldering multiple myeloma (SMM). The progression from MGUS/SMM to MM can be determined by an increase of genetic alterations. Patients with this disease typically present infiltration of clonal plasma cells into the bone marrow and monoclonal protein into the serum. Genetic alterations found in bone marrow biopsies have also been found by cell-free circulating tumor DNA (cfDNA) and circulating plasma cells (CPCs), purified from liquid biopsies (LBs). LBs represent a minimally-invasive alternative to tissue biopsies. The aim of my project is to investigate the increase of genetic mutations load on LBs of patients with different disease stages.

**Methods**

Collection of biopsies samples from patients affected by MGUS, SMM, MM.

Plasma and CPCs separation from blood samples.

Extraction of DNA from biopsies samples.

Optimization of specific protocols for separation and extraction.

**Results**

During this first year, I focused my activity on the collection and storage of samples. I have collected over 1800 samples including blood samples and bone marrow biopsies. From each sample collected, the plasma was separated for the detection of cfDNA while the Buffy coat was taken for the detection of CPCs, implementing and modifying the protocols already present in the literature. A database has been created to annotate specific diagnostic parameters to select a good cohort of samples for the next step of investigation of the mutational profile that includes NGS sequencing. 170 MM, 190 MGUS, 20 SMM have already been selected, the identification and selection of many other samples are ongoing. The preliminary DNA extraction was performed on some initial samples.

- Abstracts and participation in congresses and courses: authors, title of the presentation, name and dates of the congress
  - Participation in Congress – Virtual Edition: “XXIII Congresso Nazionale SIGU”; Authors: Biomedia srl – SIGU (Società Italiana di Genetica Umana); 11-13 November 2020
  - Participation in the training course – Virtual Edition: “Scenario e prospettive del nuovo programma europeo per la ricerca e l’innovazione”; Authors: APRE (Agenzia per la Promozione della Ricerca Europea); 11-12 January 2021
  - Participation in a conference – Virtual Edition: “Ricerca Biomedica tra diritto alla salute e valori costituzionali”; Authors: University of Cagliari - Department of Biomedical Sciences; 25 January 2021
  - Participation in Soft skills courses for PhD at the University of Siena – Virtual Edition:
    - “Diritto dei brevetti”; Authors: Vittorio Santoro; 10 e 13 May 2021
    - “Diritto dei brevetti”; Authors: Alessandro Palmieri; 11 e 12 May 2021
    - “Spin off e Start up della ricerca: concetti introduttivi e presupposti per la nascita di un’impresa”; Authors: Lorenzo Zanni; 17 May 2021
    - “Creating value from large archive and big data”; Authors: Laura Neri; 25 May 2021
    - “How to exploit your research idea: a quick introduction to EU project design and management”; Authors: Donata Franzi; 27 May 2021
    - “I rapporti tra scienza e società: tra persistenze e cambiamenti”; Authors: Agnes Allansdottir; 31 May 2021
    - “Analisi della risposta immunitaria alla vaccinazione e all’infezione: dalla fase di sperimentazione a quella di analisi dati tramite un approccio di systems biology”; Authors: Annalisa Ciabattini ed Elena Pettini; 8 June 2021
    - “Spin off e Start up della ricerca: L’ecosistema di innovazione a supporto delle start up”; Authors: Francesco Maria Senatore; 15 June 2021
    - “Lavoro editoriale per l’editoria scolastica e universitaria (scienze e lettere)”; Authors: Alessandro Mongatti; 17 June 2021
  - Participation in the English course: “Inglese: Corsi Generali A.A. 2020-21”; Authors: CLI (Centro Linguistico, Università di Pisa); 60 hours from March to June 2021
- Scientific publications, authors, title of publication, name and number of the journal, year of publication
- Possible stays in other Italian or foreign laboratories
  - Scientific Research and Training activities: “Monitoring, protection and safeguard of *Caretta caretta* sea turtle nests in Calabria”; Organization: Caretta Calabria Conservation ONLUS; Activities carried out as an activist and team leader: Protection of the nest of loggerhead sea turtle and study on the genetic characterization of nesting females of *Caretta caretta*; From 7 August 2021 to 22 August 2021.