

Al collegio docenti del Dottorato in Medicina Molecolare

Dott. Giannini Marco

Ciclo XXXVII Tutor: Prof.ssa Betti Giusti

Dottorato di ricerca a tema vincolato: Medicina molecolare in ambito cardiovascolare e aterotrombotico: utilizzo di tecnologie “omiche” e approcci di biomedicina computazionale (gestione di BigData) per la comprensione delle basi genetico-molecolari e lo sviluppo di strategie diagnostiche personalizzate

Attività scientifica svolta nel 2° anno di Dottorato, Anno Accademico 2022/2023

I attended my second year of PhD at the Advanced Molecular-Genetic Laboratory of Center for Atherothrombotic Diseases, Dpt. Experimental and Clinical Medicine, University of Florence.

Part of my year focused on continuing the analysis already begun and described in the first year's report (identification of genetic variants and/or genetic profiles associated with severity of COVID19 disease); a further targeted Next Generation Sequencing (NGS) was performed on the same cohort of n=40 patients hospitalized at AOU Careggi due to SARS-CoV-2 infection. The genetic analysis included a sequencing panel of 7 genes selected based on their involvement in cellular mechanisms that determine the anchoring, entry and life cycle of the SARS-CoV-2 virus in host cell. Preliminary data obtained seem to indicate that genetic variability may help modulate susceptibility to viral infections. We are completing a database of clinical data from more than 1,000 COVID patients in anticipation of selecting additional samples for NGS analysis next year.

Moreover, I took part in a study about VITT (Vaccine-induced Immune Thrombotic Thrombocytopenia), a prothrombotic syndrome observed in a small number of individuals who received the adenoviral vector-based vaccine Vaxzevria. We performed a WES analysis on 6 patients to identify genetic variants associated with VITT; this work resulted in an article submitted on October 04, 2023 and awaiting revision.

In recent months, we have begun a study regarding the genetic component underlying BAV (Bicuspid Aortic Valve), the most common congenital heart defect in humans, with an incidence ranging from 0.5% to 2.0% in the general population. In particular, we are analyzing by NGS technology, using a sequencing panel of 102 genes involved in aortopathies, Marfan syndrome and related pathologies, samples from about 50 BAV patients to identify genetic variants implicated in BAV disease.

Pubblicazioni scientifiche

- (Submitted 04/10/2023) Betti Giusti, Elena Sticchi, Tommaso Capezzuoli, Rebecca Orsi, Lapo Squillantini, **Marco Giannini**, Samuele Suraci, Angela Rogolino, Francesca Cesari, Martina Berteotti, Anna Maria Gori, Elena Lotti, Rossella Marcucci - Whole exome sequencing in vaccine-induced thrombotic thrombocytopenia (VITT) – Rivista Thrombosis and Haemostasis-TH-23-10-0432

Partecipazione a webinar, seminari e congressi:

- XXVII° Congresso Nazionale Siset - Perugia, 2/5 novembre 2022
- Corso di Formazione specifica in materia di Sicurezza sul lavoro (11, 18 novembre 2022): modulo “Rischi comuni a tutte le mansioni”, modulo “Rischi chimici e cancerogeni”, modulo “Rischi biologici”, modulo “Rischi Attrezzature, uso DPI e Rischi derivanti da CEM e ROA” – UNIFI
- 2nd Meeting Translational research on stroke (with Open Discussions on NIMBLE Results) – Firenze, 25 novembre 2022
- 36° Congresso Nazionale SISA - Roma 27-29 novembre 2022
- Il ruolo degli anticoagulanti iniettivi nel TEV: confronto con gli esperti – Firenze, 2 dicembre 2022
- Giornata della ricerca DMSC dal titolo “Il DMSC presenta i giovani” – Firenze, 22 dicembre 2022
- Dottorato in Medicina Molecolare, PhD Day – Firenze, 17 gennaio 2023
- Corso di Bioinformatica “Computational Systems Biology” tenuto dal Dr. Roberto Semeraro (13, 20 gennaio, 3, 17 febbraio 2023) – Dipartimento di Medicina Sperimentale e Clinica, UNIFI
- Il DMSC dalla parte delle donne, Giornata Internazionale della Donna – Firenze, 8 marzo
- Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023
- Seminario DENOThe “Molecular chaperones – cellular machineries of protein folding and proteostasis”, Prof. F. Ulrich Hartl, Department of Cellular Biochemistry, Max Planck Institute of Biochemistry, Martinsried, Germany – Firenze, 29 giugno 2023

Abstracts:

- **Marco Giannini**, Elena Sticchi, Tommaso Capezzuoli, Rosina De Cario, Samuele Suraci, Giulia Cassioli, Sara Neroni, Angela Antonietta Rogolino, Martina Berteotti, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Identification of rare coagulation variants and screening of thrombophilic polymorphisms (f2 rs1799963, f5 rs6025) in COVID-19 patients. CORSO NAZIONALE Siset PERUGIA 2022, 02/11-05/11 2022.

-Giulia Barbieri, Tommaso Capezzuoli, **Marco Giannini**, Giulia Cassioli, Francesca Cesari, Rossella Marcucci, Anna Maria Gori, Betti Giusti, Elena Sticchi. Lpa kringle iv type 2 detection: qpcr or ddpcr? CORSO NAZIONALE Siset PERUGIA 2022, 02/11-05/11 2022

-Rosina De Cario, Elena Sticchi, Ada Kura, Tommaso Capezzuoli, **Marco Giannini**, Giulia Cassioli, Giulia Barbieri, Samuele Suraci, Anna Maria Gori, Martina Berteotti, Angela Antonietta Rogolino, Rossella Marcucci, Betti Giusti. Individual susceptibility to sars-cov-2 infection: role of the host's virus entry machinery genetic profiles. CORSO NAZIONALE Siset PERUGIA 2022, 02/11-05/11 2022.

-Angela Rogolino, Francesca Cesari, Anna Maria Gori, Betti Giusti, Bandinelli Brunella, Elena Sticchi, Alessia Bertelli, **Marco Giannini**, Daniela Poli, Elena Lotti, Lucia Mannini, Rossella Marcucci. Vaccine-induced thrombotic thrombocytopenia (VITT): evaluation of immunologic and functional tests on an Italian case series. CORSO NAZIONALE Siset PERUGIA 2022, 02/11-05/11 2022.

-Tommaso Capezzuoli, Rosina De Cario, Elena Sticchi, **Marco Giannini**, Rebecca Orsi, Lapo Squillantini, Samuele Suraci, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Vaccine induced thrombotic thrombocytopenia: evaluation of genetic susceptibility through whole exome sequencing. CORSO NAZIONALE Siset PERUGIA 2022, 02/11-05/11 2022.

-Giulia Barbieri, Tommaso Capezzuoli, **Marco Giannini**, Giulia Cassioli, Francesca Cesari, Rossella Marcucci, Anna Maria Gori, Betti Giusti, Elena Sticchi - Genetic characterization of lipoprotein(a) kringle IV type 2 repeat polymorphism: comparison between digital droplet and real-time PCR - 36° Congresso Nazionale SISA - Roma 27-29 novembre 2022

-**Marco Giannini**, Elena Sticchi, Tommaso Capezzuoli, Rosina De Cario, Samuele Suraci, Giulia Cassioli, Sara Neroni, Angela Antonietta Rogolino, Martina Berteotti, Anna Maria Gori, Rossella Marcucci, Betti Giusti- screening of polymorphisms f2 rs1799963, f5 rs6025 and identification of rare coagulation variants in covid-19 patients- 36° Congresso Nazionale SISA - Roma 27-29 novembre 2022

-**Marco Giannini**, Elena Sticchi, Tommaso Capezzuoli, Rosina De Cario, Lapo Squillantini, Rebecca Orsi, Samuele Suraci, Giulia Cassioli, Sara Neroni, Giulia Barbieri, Angela Antonietta Rogolino, Martina Berteotti, Anna Maria Gori, Rossella Marcucci, Betti Giusti. Identification of rare coagulation variants and screening of polymorphisms F2 rs1799963 and F5 rs6025 in COVID-19 patients. Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart, Rimini, 16-18 aprile 2023

-Tommaso Capezzuoli, Rosina De Caro, Elena Sticchi, **Marco Giannini**, Rebecca Orsi, Lapo Squillantini, Giulia Cassioli, Giulia Barbieri, Samuele Suraci, Anna Maria Gori, Rossella Marcucci, Betti Giusti – Vaccine Induced Thrombotic Thrombocytopenia (VITT): Evaluation of Genetic Susceptibility through WES - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023

-Lapo Squillantini, Elena Sticchi, Tommaso Capezzuoli, **Marco Giannini**, Giulia Cassioli, Giulia Barbieri, Samuele Suraci, Anna Maria Gori, Martina Berteotti, Angela Antonietta Rogolino, Rosina De Caro, Ada Kura, Rossella Marcucci, B. Giusti - Individual Vulnerability to SARS-COV-2 infection: genetic profiles and the role of the host's virus entry machine - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023

-Giulia Barbieri, Anna Maria Gori, Francesca Cesari, Elena Sticchi, Angela Antonietta Rogolino, Rebecca Orsi, Tommaso Capezzuoli, Giulia Cassioli, **Marco Giannini**, Lapo Squillantini, Alessia Bertelli, Raffaele De Caterina, Betti Giusti, Rossella Marcucci - Edoxaban Effects On Platelet Function: In Vitro And Ex Vivo Studies - Spring Meeting Giovani Ricercatori, Basic and clinical research: until grant let us apart – Rimini, 16-18 aprile 2023