UNIVERSITÀ DI SIENA
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DIPARTIMENTO DI MEDICINA MOLECOLARE E DELLO SVILUPPO

Università degli Studi di Siena
Dottorato di Ricerca in Medicina Molecolare

Scheda Relazione Annuale Dottorando
(da redigere in lingua inglese)

Firenze, 19/09/2024

Al Collegio Docenti del Dottorato in Medicina Molecolare

dott**. Lapo Squillantini**

ciclo **XXXVIII** tutor **Dott.ssa Elena Sticchi**

Attività scientifica svolta nel 2° anno di Dottorato, Anno Accademico **2023/2024**

During this academic year, I have focused my research activities on the search for molecular genetic markers in heritable connective tissue disorders. In particular, I have contributed to a project of my group related to the study of the bicuspid aortic valve, where a panel of 102 genes related to the disease was sequenced on a cohort of 100 subjects; I also worked on setting up an SPSS database containing the genetic and clinical data of the subjects followed at our regional reference center for Marfan disease and related diseases, which to date includes the data of 396 subjects on whom we are carrying out genotype/phenotype correlation studies.
The methods used to carry out my project include nucleic acid extractions, PCR and real-time PCR, Targeted Next Generation Sequencing (NGS) approach of at least 17 genes associated with aortopathy (ACTA2, CBS, COL3A1, ELN, FBN1, FBN2, LTBP3, MYH11, MYLK, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGBR1, TGFBR2) performed thought SureSelectQXT library preparation kit (Agilent Technologies) on MySeq Illumina platform, bioinformatic analysis of the data obtained (I am currently following the 'advanced technologies in bioinformatics' course at the Golinelli Foundation to increase my knowledge in this field) and Sanger sequencing to validate the identified variants.
The results I have obtained so far focus on a case study of patients with pathogenic variants or likely pathogenic variants on the SMAD3 gene. Our case studies are one of the most extensive compared to the literature, with six index cases and 16 affected subjects. Our analyses highlight several cardiovascular characteristics in some cases index and relatives affected. This complements and supports the latest literature and may have an impact on the risk stratification and follow-up of these subjects. Based on these data, I will present an oral communication to the SISET congress from 6 to 9 November in Rome, and a paper will also be processed.

• Abstracts e partecipazione a congressi e corsi: autori, titolo della presentazione, nome e date del congresso

- Orsi R, Giusti B, Sticchi E, Capezzuoli T, **Squillantini L,** Giannini M, Suraci S, Rogolino A, Cesari F, Berteotti M,Gori AM, Lotti E, Marcucci R. WHOLE EXOME SEQUENCING IN VACCINE-INDUCED THROMBOTIC THROMBOCYTOPENIA (VITT). Spazio Giovani, XXXVII Congresso Nazionale della Società Italiana per lo Studio dell'Arteriosclerosi (SISA), Napoli, 26-28 novembre 2023.

- Sticchi E, Giusti B, Capezzuoli T, Orsi R, **Squillantini L**, Giannini M, Suraci S, Rogolino AA, Cesari F, Berteotti M, Gori AM, Lotti E, Marcucci R. WHOLE EXOME SEQUENCING IN VACCINE-INDUCED THROMBOTIC THROMBOCYTOPENIA (VITT). XXII Riunione Gruppo di Studio delle Piastrine, Gubbio, 1-3 ottobre 2023.

- MUTATIONS IN SMAD3 GENE: RELATIONSHIP WITH CARDIOVASCULAR FEATURES **Squillantini L**, Sticchi E, Capezzuoli T, Giannini M, Barbieri G, kura A, Suraci S, Rogolino A, Gensini F, Nistri S, Pepe G, Gori AM, Berteotti M, Marcucci R, Giusti B. Roma, 6/9 novembre 2024

• Pubblicazioni scientifiche autori, titolo della pubblicazione, nome e numero della rivista, anno di pubblicazione

- Betti Giusti, Elena Sticchi, Tommaso Capezzuoli, Rebecca Orsi, **Lapo Squillantini**, Marco Giannini, Samuele Suraci, Angela Antonietta Rogolino, Francesca Cesari, Martina Berteotti, Anna Maria Gori, Elena Lotti, Rossella Marcucci. Whole Exome Sequencing in Vaccine-Induced Thrombotic Thrombocytopenia (VITT), BioMed Research International, 2024

• Eventuali soggiorni in altri laboratori italiani o esteri