

Dott. Samuele Suraci

Ciclo XXXIV Tutor Prof.ssa Betti Giusti

Attività scientifica svolta nel 2° anno di Dottorato, Anno Accademico 2019/2020

The research activity was carried out at the Advanced Molecular-Genetics Laboratory of the Atherothrombotic Diseases Centre, Department of Experimental and Clinical Medicine, University of Florence. This activity is part of an extensive research project aimed to study the clinical aspects and genetic/molecular bases of complex cardiovascular traits through “omics” and innovative computational approaches.

Object of this second year of PhD was the optimising for clinical use of the bioinformatic pipeline I developed during the first year of PhD and its application for mutational analysis through "targeted" high throughput sequencing (HTS) of large genes panels.

One of these panel addresses genes involved, according to most recent literature data, in pathogenesis of Bicuspid Aortic Valve (BAV), syndromic and non-syndromic aortopathy, and development and remodelling of large vessels and heart valves.

The aim of the work I carried out during this year is to define a set of parameters and an algorithm that allow to confidently exclude false positive and retain only the actual variants called from HTS experiments.

The evaluation of this panel in our cohort of BAV patients allowed us to reach interesting results. We are finalizing a paper for an international scientific journal. In this study, besides confirming association of BAV with *NOTCH1* gene, we observed new genes whose implication in the pathogenesis of the disease was not previously hypothesized. Our data support the hypothesis of a polygenic nature of BAV, suggesting that mutations in different genes can lead to a similar clinical phenotype. We are currently evaluating, increasing the number of subjects, a possible association of variants in different genes or different types of variants, with peculiar characteristics (presence of aortic dilation, aortic dilation at different levels) or different complications (calcification, stenosis, type of BAV) of the disease.

I adapted this pipeline to work also with different gene panels and library preparation kits.

I was also involved in carrying out a bioinformatic analysis to evaluate global RNA expression profiles from cerebral thrombi obtained during thrombectomy treatment, and from venous peripheral blood in patients with acute ischemic stroke, using Affymetrix technology.

Finally, I carried out a bioinformatic analysis to evaluate through Small RNA Sequencing the presence of Torque teno virus microRNA nucleic acid motif in cerebrospinal fluid in subjects under assessment of neurological diseases. Four CSF samples positive for all three TTV microRNAs were selected from a total of 93 samples, obtained from patients with meningitis/encephalitis disorders during their viral diagnostic assessment. Total RNA was isolated from extracellular small vesicles

obtained from 250 µl CSF sample processed with Exosomal RNA Purification kit (NorgenBiotek Corp., ON, Canada) and following the manufacturer's protocol. Libraries were prepared using the special "Small RNA library prep # LX05896" kit developed by Lexogen. I developed a specific bioinformatic pipeline to check the presence of mature ttv microRNA compatible sequences in the samples. Briefly: adapters were trimmed using BBDuck.sh from BBTools. No length filters were applied. A custom FASTA file, containing previously described TTV mature microRNA sequences and all known human mature microRNA sequences from miRBase 22.1, was used as reference. Trimmed reads were aligned to the reference using Bowtie allowing no mismatch. Reads mapping to each mature miRNA were counted and normalised to Reads per Million (RPM) using a custom script in R. Reads compatible with TTV microRNA were found in all CSF samples except for sequence of one TTV miRNA in one CSF sample.

Articoli su rivista scientifica

- SANGER VALIDATION OF HIGH-THROUGHPUT SEQUENCING IN GENETIC DIAGNOSIS: STILL THE BEST PRACTICE?

Rosina De Carlo - Ada Kura - Samuele Suraci - Alberto Magi - Andrea Volta - Rossella Marcucci - Anna Maria Gori - Guglielmina Pepe - Betti Giusti - Elena Sticchi (Submitted to Frontiers in Genetics 2020)

- TORQUETENOVIRUS MICRORNA DETECTION IN CEREBROSPINAL FLUIDS OF PATIENTS WITH NEUROLOGICAL PATHOLOGIES

Simone Giannecchini - Lorenzo Neri - Pier Giorgio Spezia - Samuele Suraci - Lisa Macera - Stefano Scribano - Betti Giusti - Daniele Focosi - Fabrizio Maggi (Submitted to Journal of Clinical Virology 2020)

- GENETIC BASES OF BICUSPID AORTIC VALVE: CONFIRMATORY AND NOVEL EVIDENCES

Elena Sticchi*, Rosina De Carlo*, Samuele Suraci, Alberto Magi, Maria Pia Fugazzaro, Stefania, Colonna, Anna Maria Gori, Rossella Marcucci, Stefano Nistri, Guglielmina Pepe, Betti Giusti (in preparation)

Abstracts e partecipazione a congressi

33° CONGRESSO NAZIONALE BOLOGNA S.I.S.A. 24-26 NOVEMBRE 2019, ROMA:

- THROMBUS AND PERIPHERAL VENOUS BLOOD GLOBAL GENE EXPRESSION PROFILES IN ACUTE ISCHEMIC STROKE: COMPLEMENTARY OR PARALLEL REALITIES? (poster)

A. Kura, A. Sodero E. Sticchi, A. Sereni, S. Galora, A. Magi, S. Suraci, R. De Carlo, A. Consoli, A. Rosi, S. Nappini L. Renieri N. Limbucci, B. Piccardi, C. Sarti, D. Inzitari, S. Mangiafico, A.M. Gori, R. Marcucci, B. Giusti

- GENETIC DETERMINANTS OF FAMILIAL HYPERCHOLESTEROLEMIA: WHAT ELSE BEYOND LDLR GENE? (Comunicazione Orale)

A. Kura, E. Sticchi, A. Volta, R. De Carlo, A. Murri, A. Magi, S. Suraci A.M. Gori, R. Marcucci, B. Giusti

- SANGER VALIDATION OF HIGHTHROUGHPUT SEQUENCING IN GENETIC DIAGNOSIS: STILL THE BEST PRACTICE?

R. De Carlo, E. Sticchi, A. Kura, S. Suraci, S. Galora, A. Magi, A. Volta, R. Marcucci, S. Nistri, G. Pepe B. Giusti

- ROLE OF TALMUD POLYGENIC SCORE IN A TUSCANY COHORT OF FAMILIAL HYPERCHOLESTEROLEMIA WITH AND WITHOUT LDLR GENE MUTATIONS

E. Sticchi, A. Kura, A. Volta, R. De Carlo, A. Murri, A. Magi, S. Suraci, A.M. Gori, R. Marcucci, B. Giusti

XXVI CONGRESSO NAZIONALE SISET BAVENO (VB) 5/7 Novembre 2020 (ACCETTATI)

- FAMILIAL HYPERCHOLESTEROLEMIA: INSIGHT INTO OTHER MAJOR GENETIC DETERMINANTS BEYOND LDLR GENE

Elena Sticchi - Ada Kura - Andrea Volta - Rosina De Carlo - Alberto Magi - Samuele Suraci - Anna Maria Gori - Rossella Marcucci - Betti Giusti

- COMMON GENETIC VARIANTS IN THE MOLECULAR CHARACTERIZATION OF FAMILIAL HYPERCHOLESTEROLEMIA: THE ROLE OF TALMUD POLYGENIC SCORE

Elena Sticchi - Ada Kura - Andrea Volta - Rosina De Carlo - Alessandra Murri - Angela Rogolino - Brunella Bandinelli - Alberto Magi - Samuele Suraci - Anna Maria Gori - Rossella Marcucci - Betti Giusti

- GENETIC BASES OF BICUSPID AORTIC VALVE: CONFIRMATORY AND NOVEL EVIDENCES

Elena Sticchi - Rosina De Carlo - Alberto Magi - Andrea Volta - Ada Kura - Samuele Suraci - Maria Pia Fugazzaro - Stefania Colonna - Anna Maria Gori - Rossella Marcucci - Stefano Nistri - Guglielmina Pepe - Betti Giusti

- CEREBRAL THROMBI AND PERIPHERAL VENOUS BLOOD TRANSCRIPTOMICS: A PROMISING RESOURCE TO COMPREHEND PATHOPHYSIOLOGY AND OUTCOMES IN

ACUTE ISCHEMIC STROKE (COMUNICAZIONI ORALI SELEZIONATE - Sessione Plenaria
1" dedicata alle Migliori Comunicazioni Orali)

Ada Kura - Alessandro Soderò - Elena Sticchi - Alberto Magi - Samuele Suraci - Rosina De Carlo -
Arturo Consoli - Andrea Rosi - Sergio Nappini - Leonardo Renieri - Nicola Limbucci - Benedetta
Piccardi - Francesco Arba - Cristina Sarti - Domenico Inzitari - Salvatore Mangiafico - Anna Maria
Gori - Rossella Marcucci - Betti Giusti

- DETERMINANTS OF ONSET AND POOR OUTCOMES IN ACUTE ISCHEMIC STROKE:
ROLE OF BIOLOGICAL MARKERS IN RISK STUDY

Ada Kura - Alessandro Soderò - Elena Sticchi - Alberto Magi - Samuele Suraci - Rosina De Carlo -
Arturo Consoli - Andrea Rosi - Sergio Nappini - Leonardo Renieri - Nicola Limbucci - Benedetta
Piccardi - Francesco Arba - Cristina Sarti - Domenico Inzitari - Salvatore Mangiafico - Rossella
Marcucci Betti Giusti - Anna Maria Gori

- IS SANGER VALIDATION OF HIGH-THROUGHPUT SEQUENCING STILL MANDATORY
IN GENETIC DIAGNOSIS?

Rosina De Carlo, Elena Sticchi, Ada Kura, Samuele Suraci, Alberto Magi, Andrea Volta, Anna
Maria Gori, Rossella Marcucci, Guglielmina Pepe, Betti Giusti

- ENDOTHELIAL DYSFUNCTION, THROMBIN GENERATION AND BRINOLYSIS IN
COVID-19 PATIENTS

Elena Sticchi - Anna Maria Gori - Betti Giusti - Ada Kura - Rosina De Carlo - Brunella Bandinelli -
Angela Rogolino - Daniela Poli - Lucia Mannini - Samuele Suraci - Elena Lotti - Rossella Marcucci

34° Congresso Nazionale S.I.S.A. Virtual edition, 22-24 Novembre 2020 (SOTTOMESSI)

- BIOHUMORAL MARKERS OF ACUTE ISCHEMIC STROKE: THE RISK STUDY

Ada Kura - Alessandro Soderò - Elena Sticchi - Alberto Magi - Samuele Suraci - Rosina De Carlo -
Arturo Consoli - Andrea Rosi - Sergio Nappini - Leonardo Renieri - Nicola Limbucci - Benedetta
Piccardi - Francesco Arba - Cristina Sarti - Domenico Inzitari - Salvatore Mangiafico - Rossella
Marcucci - Betti Giusti - Anna Maria Gori

- TRANSCRIPTOMICS STUDY IN CEREBRAL THROMBI AND PERIPHERAL
VENOUS BLOOD OF ACUTE ISCHEMIC STROKE PATIENTS

Ada Kura - Alessandro Soderò - Elena Sticchi - Alberto Magi - Samuele Suraci - Rosina De Carlo -
Arturo Consoli - Andrea Rosi - Sergio Nappini - Leonardo Renieri - Nicola Limbucci - Benedetta
Piccardi - Francesco Arba - Cristina Sarti - Domenico Inzitari - Salvatore Mangiafico - Anna Maria
Gori - Rossella Marcucci - Betti Giusti

- SANGER SEQUENCING DATA VS HIGH QUALITY NGS DATA: A COMPARISON OF RELIABILITY.

Rosina De Caro, Ada Kura, Samuele Suraci, Alberto Magi, Andrea Volta, Anna Maria Gori, Rossella Marcucci, Guglielmina Pepe, Betti Giusti, Elena Sticchi

- TALMUD POLYGENIC SCORE: CONTRIBUTION OF COMMON GENETIC VARIANTS IN THE MOLECULAR CHARACTERIZATION OF FAMILIAL HYPERCHOLESTEROLEMIA

Ada Kura Elena Sticchi - Andrea Volta - Rosina De Caro - Alessandra Murri - Angela Rogolino - Brunella Bandinelli - Alberto Magi - Samuele Suraci - Anna Maria Gori - Rossella Marcucci - Betti Giusti

- ENDOTHELIAL DYSFUNCTION, THROMBIN GENERATION AND FIBRINOLYSIS IN COVID-19 PATIENTS

Elena Sticchi - Anna Maria Gori - Betti Giusti - Ada Kura - Rosina De Caro - Brunella Bandinelli - Angela Rogolino - Daniela Poli - Lucia Mannini - Samuele Suraci - Elena Lotti - Rossella Marcucci

- INSIGHT INTO FAMILIAL HYPERCHOLESTEROLEMIA GENETIC DETERMINANTS BEYOND LDLR LOCUS

Elena Sticchi - Ada Kura - Andrea Volta - Rosina De Caro - Alberto Magi - Samuele Suraci - Anna Maria Gori - Rossella Marcucci - Betti Giusti

Seminari e conferenze

- DENOThe Meeting 2019 – 1/10/2019
- “La percezione pubblica della scienza: i giovani ricercatori di fronte a temi scientifici di forte interesse pubblico, politico e mediatico” 27-28/04/ 2020
- “Covid-19: come si insegue e sconfigge un virus” 28-29/05/2020
- PANDEMIA DA SARS-COV-2: ASPETTI VIRALI, EPIDEMIOLOGICI, CLINICI, IMMUNOLOGICI, E TERAPEUTICI 04/06/2020
- WEBINAR Siset HIT - trombocitopenia indotta da eparina: laboratorio e clinica Prof.ssa Rossella Marcucci 16/06/2020
- Illumina Experience 2020 16/06/2020
- Controllo Qualità in NGS: Quando qualificare fa la differenza 23/07/2020
- Dalla preparazione delle librerie all’interpretazione - Il Workflow di Agilent su casi reali Thursday, 16/07/2020

- L'importanza della LOH e della sua interpretazione nella pratica clinica Thursday, 25/06/2020
- Soluzioni di automazione in NGS per uso clinico: un caso pratico “walk away” 2/07/2020
- Webinar Siset del Dott. Marco Marietta "MODELLI DI COAGULOPATIE ACQUISITE: TIC, DIC, SIC e CAC" 08/09/2020

Complementary Skills e Corsi

- DIRITTO DEI BREVETTI - Prof. Vittorio Santoro 11 e 13/05/2020
- SPIN OFF E START UP DELLA RICERCA: IL MODELLO DI BUSINESS E I REGOLAMENTI – Prof. Niccolò Fiorini 9/06/2020
- DIRITTO DEI BREVETTI – Prof. Alessandro Palmieri 15 e 22/06/2020
- LAVORO EDITORIALE PER L'EDITORIA SCOLASTICA E UNIVERSITARIA - Dr. Alessandro Mongatti 16/06/2020
- I RAPPORTI TRA SCIENZA E SOCIETÀ TRA PERSISTENZE - Dr. Agnes Allansdottir 17/06/2020
- HOW TO EXPLOIT YOUR RESERCH IDEA: A QUICK INTRODUCTION TO EU PROJECT DESIGN AND MANAGMENT – Dr. Donata Franzi 23/06/2020
- SOFT SKILLS PER FARE RICERCA NELLE ORGANIZZAZIONI – Prof. Alessandra Romano 10/07/2020

Altre Attività

Realizzazione e produzione video lezioni pratiche del corso “**TECNICHE IN BIOTECNOLOGIE ELABORATORIO DI BIOTECNOLOGIE**”, Modulo B016876, Università di Firenze.