

CURRICULUM VITAE

Personal data

Surname and First name Frullanti Elisa
Place and Date of birth Siena – ITALY - 17/03/1982
Address Via della libertà 5B - 53040 Rapolano Terme (SI) ITALY
Telephone +39 3426200415 (personal)
+39 0577233303 (work)
E-Mail elisa.frullanti@dbm.unisi.it
Nationality Italian

Education

- 2011 – 2012 University of study of Milan, Italy
Professional continuing education course on "Medical statistics and Statistical Methods for Epidemiology".
Level in national or international classification: ISCED5
- 2007 - 2011 Open University, London (UK), affiliated research centre: Fondazione IRCCS Istituto Nazionale per lo studio e la cura dei Tumori, Milan (Italy)
PhD Degree in Cancer Genetics
Research Project: "*Genome-wide identification of functional polymorphisms modulating individual risk or prognosis of lung cancer*"
Level in national or international classification: ISCED6
- 2004 - 2006 University of study of Milan, Italy.
Degree in Molecular Biology of the Cell (final mark 110/110 *summa cum laude*).
Experimental thesis title: "*Genome-wide analysis for the identification of polymorphisms involved in lung cancer risk*".
Level in national or international classification: ISCED5
- 2001 - 2004 University of study of Siena, Italy.
First Level Degree in Biological Sciences (final mark 110/110 *summa cum laude*).
Experimental thesis titled "*Cloning and expression of mVEGFR-3 receptor*".
Level in national or international classification: ISCED5
- 1996 - 2001 High school "Scientific Lyceum G. Galilei", Siena, Italy.
Scientific Lyceum Diploma obtained with final mark 100/100.
Level in national or international classification: ISCED3

Employment

- 1 August 2016 – Onward Assistant Professor - Operative Unit of Medical Genetics, University of Siena, Viale Bracci 2 – 53100 Siena (Italy) Academic Field/Discipline: 06/A1 Medical Genetics/MED/03 Medical Genetics
- May 2012 – July 2016 Postdoctoral fellow - Operative Unit of Medical Genetics, University of Siena, Viale Bracci 2 – 53100 Siena (Italy)
- May 2011 – April 2012 Postdoctoral fellow - Research Unit "Genetics Epidemiology and Pharmacogenomics" – Department of Predictive and Preventive Medicine – Fondazione IRCCS Istituto Nazionale per lo Studio e la Cura dei Tumori Via Venezian 1 – 20133 Milan (Italy)
- July 2006 – May 2011 Research fellow - Research Unit "Genetics Epidemiology and Pharmacogenomics" –

Department of Predictive and Preventive Medicine – Fondazione IRCCS Istituto Nazionale per lo Studio e la Cura dei Tumori
Via Venezian 1 – 20133 Milan (Italy)

December 2004 – June 2006 Research Fellow - Molecular and Genetic Epidemiology Unit –
Fondazione Policlinico IRCCS Regina Elena –
Via Pace 9 – 20122 Milan (Italy)

May 2004 – September 2004 Research Fellow – Department of Molecular Biology – University of study of Siena
(Italy)

Awards

2011 - "Galilei Giovani" Young Research Scientist Award, Rotary International (District 2100 – Italy)

Technical skills and competences

- Competency on genome-wide association study analyses, and transcriptome studies to identify loci and candidate genes associated with lung tumorigenesis.
- Expertise in SNP genotyping by pyrosequencing; skills in standard molecular biology techniques for qualitative and quantitative analyses of nucleic acids: standard or real-time PCR, gene cloning, DNA sequencing through Sanger or Next-Generation Technique, etc.
- Expertise in dissecting the molecular and genetic bases of LC and other diseases following innovative "omics" approaches which integrate genomic (WES and WGS) and transcriptomic (RNAseq) data.
- Experience in data analysis using specific tools for nucleotide sequence analysis (Chromas, BLAST, Genomatix) and in whole-exome data analysis. Expertise in computational biology using browsers (NCBI, Ensembl genome, HapMap Project, etc.).
- Expertise in statistical analysis for biological data using R packages, BRB ArrayTools, Review Manager and SPSS software.
- Experience in writing projects and scientific articles and in peer-reviewing scientific papers for international journals.

Teaching Responsibilities

- Since 2013 *Module of "Medical Genetics" within the C.I. of General Pathology and Medical Genetics* 2nd year, CdL Biomedical Laboratory Techniques, University of Siena
- Since 2015 Teacher in the *Master in "Genetic-molecular Pathology"*, University of Siena
- Since 2016 Teacher in the *Specialty School in Medical Genetics*, University of Siena
- Since 2017 Teacher in the *Research Doctorate in Genetics, Oncology and Clinical Medicine (GenOMeC)*, University of Siena

Diagnostic activity

Intellectual disability: analysis of the CGH array in patients with cognitive impairment, autism and multiple congenital anomalies.

Medullary thyroid carcinoma: pRET screening in patients with sporadic and familial thyroid carcinoma.

Editorial Board

- Since 2017 Journal of Pulmonary & Respiratory Sciences (OAJPRS)
- Since 2018 Future Research in Cancer and Medicine
- Since 2018 Journal of Integrative Medicine

Affiliations to Scientific Societies and Committee

- *Società Italiana di Genetica Umana (SIGU)*
- *Società Italiana di Cancerologia (SIC)*
- *American Society of Human Genetics (ASHG)*
- *European Society of Human Genetics (ESHG)*

Participation in Editorial Committees

- 2018: Rivista Scientifica internazionale "*Future Research in Cancer and Medicine*"
- 2017: Rivista Scientifica internazionale "*Journal of Pulmonary & Respiratory Sciences (OAJPRS)*"

Patent & Technology Transfer

- From 05/02/2018 - participation in "Siena Gen Test (SGT)" (<http://sienagentest.dbm.unisi.it>) - Spin-off Universitaria

Grants

Title: *A multidisciplinary approach to study protocadherin 19: from neuronal function to the "cellular interference" pathogenic mechanism*

Funding: PRIN (Research Projects of National Relevance) - Italian Ministry of Education, Universities and Research (MIUR)

Duration: 2019-2022

Role in the project: Prof. Elisa Frullanti (Director of Research Unit-Partner)

Budget for Frullanti's Unit: 168.000 Euro

National Scientific Qualification

- SSC 05/I1 – GENETICS for Associate Professor (from 12/04/2017 to 12/04/2023)

Congress and Meeting

- 1ST World Congress on Gender-Specific Medicine, "*Men, women and medicine*", Berlin (Germany), 23-26 February 2006. Poster presentation.
- 6th Joint PhD Student Workshop, Riva del Garda (Italy), 21-23 January 2008.
- 20th Meeting of the European Association for Cancer Research (EACR), Lyon (France), 5-8 July 2008.
- 4^o meeting A.I.F.E.G.-SIGU: *Predisposizione Allo Sviluppo Di Tumori Gastrointestinali: Il Ruolo Dei Geni A Bassa Penetranza*, Villa San Fermo, Lonigo (Vicenza), 12-13 May 2009
- *Irish Association for Cancer Research Annual Meeting*, Galway (Ireland), 3-5 March 2010
- 16th Charles Heidelberger Symposium on Cancer Research, Coimbra (Portugal), 26-28 September 2010.
- 52^o Annual Meeting of the Italian Cancer Society – *LOST IN TRANSLATION: bridging the gap between cancer research and effective therapies*, Rome, 4-7 October 2010.
- *IARC Summer School in Cancer Epidemiology 2011* – International Agency for Research on Cancer - World Health Organization, Lyon (France), 27 June - 8 July 2011.
- 53^o Annual Meeting of the Italian Cancer Society – *Back to the Future: Translating Cancer Research from Bedside to Bench and Back*, Turin, 19-22 October 2011.

- Stage for usage of Dynamic Array™ integrated fluidic circuits (IFCs) technology by Fluidigm, for genotyping solution with SNPtype custom Assays – Policlinico S.Orsola – Malpighi, Bologna (Italy), 25-29 June 2012.
- 22nd Biennial Congress of the European Association for Cancer Research (EACR), Barcelona (Spain), 7-10 July 2012.
- XXVII Conferenza nell'ambito delle Conferenze dei Presidi sulle Malattie Rare: "La centralità della Toscana nella ricerca mondiale su una malattia rara: l'alcaptonuria", Azienda ospedaliera di Careggi, Florence, Italy, 3 October 2012.
- 3° Conferenza Nazionale sulla Ricerca Sanitaria, Villa Erba, Cernobbio (CO), Italy, 12-13 November 2012.
- EPIRARE Second International Workshop "Rare disease and orphan drug registries", Rome, Italy, 21-22 October 2013.
- "L'efficienza organizzativa tra strategia ed operatività: il Lean Thinking in Sanità", Siena, Italy, 22 November 2013.
- Primo concorso LEAN, Azienda Ospedaliera Universitaria Senese Siena, Italy, 10 December 2013.
- Course on "Agilent CytoGenomics 2.7" (Agilent Biotechnology), Azienda Ospedaliera Universitaria Senese, Siena, Italy, 18 March 2014.
- European Human Genetics Conference (ESHG) 2014 in conjunction with The European Meeting on Psychosocial Aspects of Genetics (EMPAG) 2014 and the Società Italiana di Genetica Umana (SIGU), Milan, Italy, May 31 – June 3, 2014.
- Life Technologies Ion Proton System operational training course (Life Technologies), Azienda Ospedaliera Universitaria Senese, Siena, Italy, 9 June 2014.
- Workshop "European Reference Networks", Istituto Superiore di Sanità, Roma, Italy, 3 July 2015.
- FAD Course "Eterogeneità genetica nei tumori ereditari: quali test proporre nella pratica clinica", Azienda Ospedaliera Universitaria Senese, Siena, Italy, 17 July 2015.
- XVIII Congress of Società Italiana di Genetica Umana (SIGU), Rimini, Italy, October 21-24, 2014.
- Course on "Agilent CytoGenomics 3.0" (Agilent Biotechnology), Azienda Ospedaliera Universitaria Senese, Siena, Italy, 5 November 2015.

Speaker at Conferences and Courses

- *The personalized inherited signature predisposing to non-small cell lung cancer in non-smokers.* XIX Congresso Nazionale SIGU 2016 - 23-26 Novembre 2016, Torino (Italia)
- *Big Data in Medical Genetics: the Genomic Medicine. Corso Soft Skills nell'ambito dei corsi trasversali nei Dottorati di ricerca - A.A. 2017/2018* 21 Febbraio 2018, Siena (Italia)

Abstracts and Posters

- **Frullanti E**, Forti S, Raimondi S, Benhamou S, Cascorbi I, Dally H, Le Marchand L, London SJ, Risch A, Spitz MR, Stucker I, Wu X, Yang P, Taioli E. *Age and gender differences in the association between MPO 463G→A and lung cancer. Results from the Genetic Susceptibility to Environmental Carcinogens pooled analysis.* 1ST World Congress on Gender-Specific Medicine, Men, women and medicine, Berlin (Germany), 23-26 February 2006. Poster presentation.
- **Frullanti E**, Galvan A, Falvella FS, Dragani TA. *Genome-Wide Identification Of Functional Polymorphisms Modulating Individual Risk Of Lung Cancer.* 6th Joint PhD Student Workshop, Riva del Garda (Italy), 21-23 January 2008. Poster presentation.

- **Frullanti E**, Galvan A, Falvella FS, Dragani TA. *Genome-Wide Identification Of Functional Polymorphisms Modulating Individual Risk Of Lung Cancer*. 20th Meeting of the European Association for Cancer Research (EACR), Lyon (France), 5-8 July 2008. Poster presentation. Published in EJC SUPPLEMENTS, 6(9):42-42. Poster presentation.
- **Frullanti E**, Galvan A, Falvella FS, Dragani TA. *Genome-wide association study in discordant sibships identifies multiple inherited susceptibility alleles linked to lung cancer*. Irish Association for Cancer Research Annual Meeting, Galway, Ireland, 3-5 March 2010. Poster Presentation.
- **E. Frullanti**, C. Berking, N. Harbeck, P. Jézéquel, A. Haugen, C. Mawrin, O. Parise Jr., H. Sasaki, N. Tsuchiya, T. A. Dragani. *FGFR4 GLY388ARG POLYMORPHISM MODULATES CANCER PATIENTS' SURVIVAL*. 16th Charles Heidelberger Symposium on Cancer Research, Coimbra, 26-28 September 2010. Poster Presentation.
- **E. Frullanti**, C. Berking, N. Harbeck, P. Jézéquel, A. Haugen, C. Mawrin, O. Parise Jr., H. Sasaki, N. Tsuchiya, T. A. Dragani. *FGFR4 GLY388ARG POLYMORPHISM MODULATES CANCER PATIENTS' SURVIVAL*. 52° Annual Meeting of the Italian Cancer Society – LOST IN TRANSLATION: bridging the gap between cancer research and effective therapies, Rome, October 4-7 2010. Poster Presentation.
- **E. Frullanti**, F. Colombo, F.S. Falvella, A. Galvan, S. Noci, L. De Cecco, M. Incarbone, L. Santambrogio, U. Pastorino, T. A. Dragani. Lung adenocarcinoma clinical stage is associated with gene expression pattern in adjacent normal lung tissue. 53° Annual Meeting of the Italian Cancer Society – Back to the Future: Translating Cancer Research from Bedside to Bench and Back, Turin, 19-22 October 2011. Poster Presentation.
- **E. Frullanti**, Falvella FS, Noci S, De Cecco L, Incarbone M, Alloiso M, Santambrogio L, Tosi D, Nosotti M, Pastorino U, Dragani TA. ECM-receptor interaction signature in normal tissue distinguishes lung adenocarcinoma patients from patients with lung metastasis. 22nd Biennial Congress of the European Association for Cancer Research (EACR), Barcelona (Spain), 7-10 July 2012. Poster Presentation. Published in EJC SUPPLEMENTS, 48: S159-S160. Poster presentation.
- **Disciglio V**, Mencarelli MA, Mucciolo M, Ndoni E, **Frullanti E**, Marozza A, Di Marco C, Lo Rizzo C, Baldassarri M, Massarelli A, Canocchi V, Anderlid BM, Metcalfe K, Le Caignec C, David A, Fryer A, Boute O, Pecile V, Battini R, Novelli A, Fichera M, Romano C, Mari F, Renieri A. Interstitial 22q13 deletions not involving SHANK3 gene: a new gene contiguous syndrome? SIGU 2012, Sorrento (Italy), 21-24 November 2012. [Oral Presentation](#).
- Fallerini C, Dosa L, Tita R, **Frullanti E**, Del Prete D, Feriozzi S, Gai G, Clementi M, La Manna A, Miglietti N, Mancini R, Mandrile G, Ghiggeri G, Piaggio G, Brancati F, Diano L, Frate E, Pinciaroli A, Giani M, Castorina P, Bresin E, Giachino D, De Marchi M, Mari F, Bruttini M, Renieri A, **Ariani F**. Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. 2014 International Workshop on Alport Syndrome, Oxford (UK), 2-3 January 2014. [Oral Presentation](#).
- **Frullanti E**, Meloni I, Bruttini M, Dosa L, Fallerini C, Mari F, Renieri A. Italian Alport National Registry and Biobank. 2014 International Workshop on Alport Syndrome, Oxford (UK), 2-3 January 2014. Poster Presentation.
- G. Livide, L. Massimino, F. Ariani, I. Meloni, **E. Frullanti**, V. Broccoli, A., Renieri. Congenital Rett syndrome: study of FoXG1 impact on adult brain. ESHG 2013, Parigi (France), 8-11 June 2013. Poster Presentation.
- G. Livide, L. Massimino, S. Amabile, A. Bartolini, **E. Frullanti**, I. Meloni, F. Ariani, V. Broccoli, A. Renieri. Sindrome di Rett congenita: studio del ruolo del gene foxg1 nel Cervello adulto. SIGU Roma, Italia. 25-28 Settembre 2013. Poster Presentation.
- **Frullanti E**, Mencarelli MA, Baldassarri M, Cetta F, Mari F, Furini S, Piu P, Dragani TA, Ariani F, Renieri A. Oligogenic germline mutations predispose to early lung adenocarcinoma in non-smokers. ESHG 2014, Milano (Italy) 31 May-3 June 2014. Poster Presentation.
- **Frullanti E**, Mencarelli MA, Cetta F, Baldassarri M, Mari F, Furini S, Piu P, Dragani TA, Ariani F, Renieri A. Whole exome sequencing approach in sib pairs identifies oligogenic germline mutations predisposing to early lung adenocarcinoma in nonsmokers. ASHG 2014, San Diego (USA). Poster Presentation.
- Meloni I, Amabile S, Landucci E, Patriarchi T, **Frullanti E**, Pinto AM, Lo Rizzo C, Ariani F, Mari F, Mencarelli MA, Hell KW, Renieri A. Contribution of imbalance of excitatory/inhibitory synaptic expression in MECP2, CDLK5 and FOXG1-related disorders. The European Human Genetics Conference 2014, Milan (Italy), May 31 - June 3, 2014. Oral presentation.

- Meloni I, Amabile S, Landucci E, Patriarchi T, **Frullanti E**, Pinto AM, Lo Rizzo C, Ariani F, Mari F, Mencarelli MA, Hell KW, Renieri A. Imbalance of excitatory/inhibitory synaptic expression in Rett syndrome iPSC-based neuronal models. The European Human Genetics Conference 2015, Glasgow, Scotland (Uk), June 6 - 9, 2015. Poster Presentation.

- Bruttini M, Baldassarri M, Fallerini C, Garosi G, **Frullanti E**, Pinto AM, Mencarelli MA, Ariani F, Renieri A. Apparent autosomal dominant inheritance in Alport syndrome due to a double hit in COL4A5 gene. International Workshop on Alport Syndrome, Göttingen, September 25-27, 2015. Poster Presentation.

- Fallerini C, Da Sacco S, Pinto AM, Furini S, Meloni I, Baldassarri M, **Frullanti E**, Ariani F, Perin L, Renieri A. Transcriptome profiling of podocytes differentiated from amniotic-fluid derived patient progenitors sheds light on Alport syndrome's pathogenesis. International Workshop on Alport Syndrome, Göttingen, September 25-27, 2015. Poster Presentation.

- Renieri A, Bruttini M, Meloni I, Baldassarri M, Mari F, Fallerini C, Ariani F, **Frullanti E**, DePalo T, Ghiggeri GM, Giani M, La Manna A, Manno C, Meroni M, Pecoraro C, Pennesi C, Peruzzi L, Riegler P, Scolari F, Massella L. Italian Alport National Registry and Biobank. International Workshop on Alport Syndrome, Göttingen, September 25-27, 2015. Poster presentation.

- Landucci E, Patriarchi T, Amabile S, **Frullanti E**, Pinto AP, Lo Rizzo C, Ariani F, Mari F, Mencarelli MA, Hell JW, Renieri A, Meloni I. Contribution of excitatory/inhibitory synaptic imbalance to MECP2, CDKL5 and FOXP1 related disorders. 17th International workshop on Fragile X and other Early-Onset Cognitive Disorders. Strasbourg (France), September 27-30, 2015. Oral Presentation.

- Pinto AM, Da Sacco S, Fallerini C, Furini S, Meloni I, Baldassarri M, **Frullanti E**, Ariani F, Perin L, Renieri A. Learning pathophysiology of Alport syndrome from RNA-seq of podocytes differentiated from amniotic-fluid derived patient progenitors. American Society of Human Genetics 65th Annual Meeting 2015, Baltimore (Maryland, USA), October 6-10, 2015. Poster Presentation.

- Pinto AM, Da Sacco S, Fallerini C, Furini S, Meloni I, Baldassarri M, **Frullanti E**, Ariani F, Perin L, Renieri A, Mari F. L'analisi di RNAseq di podociti indotti da amniociti Alport mostra una riduzione generalizzata dei geni della matrice extracellulare e una sovraespressione di citochine infiammatorie. XVIII Congresso Nazionale della Società Italiana di Genetica Umana (SIGU), Rimini, 21-24 October 2015. Oral Presentation.

- Ariani F, Bianciardi L, Landucci E, Imperatore V, Lo Rizzo C, Bizzarri V, Mencarelli MA, **Frullanti E**, Renieri A, Meloni I. FOXP1 mutated iPSCs-derived neurons are prone to premature differentiation and show Wnt and TGF-beta signaling pathway alterations. XVIII Congresso Nazionale della Società Italiana di Genetica Umana (SIGU), Rimini, 21-24 October 2015. Poster Presentation.

- Meloni I, Landucci E, Bianciardi L, Amabile S, Furini S, Vaccarino F, Imperatore V, Lo Rizzo C, Mencarelli MA, **Frullanti E**, Ariani F, Renieri A. RNA sequencing analysis in iPSCs derived Rett neurons. 4° EuroRett meeting 2015, Rome, October 30-November 1, 2015. Oral Presentation.

- Pinto A, E. Landucci, L. Bianciardi, S. Daga, **E. Frullanti**, M. Brindisi, S. Butini, V. Imperatore, F. Ariani, S. Brogi, G. Campiani, A. Renieri, I. Meloni. Common morphological and transcriptome changes in Rett spectrum disorders justify a shared therapeutic approach. The European Human Genetics Conference (ESHG) 2016, Barcelona (Spain), May 21-24, 2016. Oral Presentation.

- Fallerini C, M. Baldassari, **E. Frullanti**, M. Mencarelli, A. La Manna, G. Garosi, D. Del Prete, A. Pinto, F. Ariani, F. Mari, A. Renieri. The coexistence of two causative mutations leads to reconsider Alport syndrome pattern of inheritance. The European Human Genetics Conference (ESHG) 2016, Barcelona (Spain), May 21-24, 2016. Poster presentation.

- E. Landucci, L. Bianciardi, S. Daga, A.M. Pinto, **E. Frullanti**, M. Brindisi, S. Butini, V. Imperatore, F. Ariani, S. Brogi, G. Campiani, A. Renieri, I. Meloni. Shared therapeutic approaches are justified by common morphological and transcriptome changes in Rett spectrum disorders. ASHG 2016, Vancouver (Canada), October 18-22, 2016. Oral presentation.

- **E. Frullanti**, C. Fallerini, M. Baldassarri, M. Ghisalberti, C. Bellan, F. Cetta, S. Furini, P. Paladini, G. Gotti, F. Ariani, A. Renieri. The personalized inherited signature predisposing to non-small cell lung cancer in non-smokers. SIGU 2016, Torino (Italy), November 23-26, 2016. Oral presentation.

- E. Landucci, L. Bianciardi, S. Daga, A.M. Pinto, **E. Frullanti**, M. Brindisi, S. Butini, V. Imperatore, F. Ariani, S. Brogi, G. Campiani, A. Renieri, I. Meloni. L'analisi del profilo trascrizionale dei neuroni ottenuti da iPSCs di

pazienti affette da sindrome di Rett rivela alterazioni dei circuiti GABAergici e del network neuronale. SIGU 2016, Torino (Italy), November 23-26, 2016. Oral presentation.

- **Frullanti E**, Fallerini C, Baldassarri M, Ghisalberti M, Bellan C, Cetta F, Furini S, Paladini P, Gotti G, Ariani F, Renieri A. Omic approach in non-smokers with adenocarcinoma pinpoints to germline susceptibility and personalized medicine. ESHG 2017, Copenhagen (Denmark), May 27-30, 2017. Poster Presentation.

- Daga S, Baldassarri M, Lo Rizzo C, Fallerini C, Imperatore V, Longo I, **Frullanti E**, Ariani F, Mencarelli MA, Mari F, Pinto AM, Renieri A. Podocytes differentiated from urine renal precursor as a tool for Alport syndrome diagnosis and for assessing therapeutic strategies based on patient-derived cells. ESHG 2017, Copenhagen (Denmark), May 27-30, 2017. Oral presentation.

- Pinto AM, Daga S, Baldassarri M, Lo Rizzo C, Fallerini C, Imperatore V, Longo I, **Frullanti E**, Massella L, Pecoraro C, Garosi G, Ariani F, Mencarelli MA, Mari F, Renieri A. Urine-derived podocytes-like cells: from a diagnostic to a CRISPR/Cas9 gene therapy perspective in Alport syndrome. ASHG, Orlando (USA), 17-21 October 2017. Poster presentation.

- Papa FT, A.M. Pinto, **E. Frullanti**, I. Meloni, R. Tita, R. Caselli, C. Fallerini, D. Lopergolo, M.A. Mencarelli, M. Bocchia, Gozzetti, A. Renieri. A. Low-level TP53 mutational load antecedes clonal expansion in Chronic Lymphocytic Leukemia. SIGU 2017, Napoli (Italy), November 15-18, 2017. Poster presentation.

- S. Daga, F. Donati, M. Baldassarri, C. Lo Rizzo, C. Fallerini, E. Landucci, V. Imperatore, I. Longo, **E. Frullanti**, L. Massella, C. Pecoraro, G. Garosi, F. Ariani, M. A. Mencarelli, F. Mari, M. Doria, A. Auricchio, S. Conticello, A. Renieri, A. M. Pinto. CRISPR/Cas9 engineering approach on urine derived podocyteslineage cells: a new therapeutic perspective in treatment of ATS. SIGU 2017, Napoli (Italy), November 15-18, 2017. Oral presentation.

- A. Currò, A. M. Pinto, F. Mari, **E. Frullanti**, V. Imperatore, D. Lopergolo, M. A. Mencarelli, A. Renieri, C. Lo Rizzo. Re-reading parents' exome for solving clinical issues. SIGU 2017, Napoli (Italy), November 15-18, 2017. Oral presentation.

Book Chapters

1- **Frullanti E** and Renieri A. "Riorganizzazione dell'attività di Genetica Medica relativa ai test genetici per malattie rare in ottica LEAN" in Lean Thinking in Sanità di Bianciardi C, Bracci L, Burroni L, Guercini J, Società Editrice Esculapio, Bologna, Italy, 2014.

Publications

1. Galvan A, Falvella FS, Spinola M, **Frullanti E**, Leoni V, Noci S, Zolin A, Spada E, Milani S, Pastorino U, Incarbone M, Santambrogio L, Gonzalez Neira A, Dragani TA. *Polygenic model with common variants may predict lung adenocarcinoma risk in humans*. Int J Cancer. 2008 Nov 15;123(10):2327-30
IF (2008): 4.734
2. Falvella FS, **Frullanti E**, Galvan A, Spinola M, Noci S, De Cecco L, Nosotti M, Santambrogio L, Incarbone M, Alloisio M, Calabrò E, Pastorino U, Skaug V, Haugen A, Taioli E, Dragani TA. *FGFR4 Gly388Arg polymorphism may affect the clinical stage of patients with lung cancer by modulating the transcriptional profile of normal lung*. Int J Cancer. 2009 Feb 3;124(12):2880-2885.
IF (2009): 4.722
3. Falvella FS, Galvan A, **Frullanti E**, Spinola M, Calabrò E, Carbone A, Incarbone M, Santambrogio L, Pastorino U, Dragani TA. *Transcription deregulation at the 15q25 locus in association with lung adenocarcinoma risk*. Clin Cancer Res. 2009 Mar 1;15(5):1837-42.
IF (2009): 6.747
4. Falvella FS, Galvan A, **Frullanti E**, Dragani TA. *Reply to the Letter to the Editor from Wang: Variants weakly correlated with CHRNA5 D398N polymorphism should be considered in transcriptional deregulation at the 15q25 locus associated with lung cancer risk*. Clin Cancer Res. 2009 Mar 1;15(5):1837-42.
IF (2009): 6.747

5. Galvan A, Falvella FS, **Frullanti E**, Spinola M, Pastorino U, Neira AG, Dragani TA. *Genome-wide association study in discordant sibships identifies multiple inherited susceptibility alleles linked to lung cancer*. *Carcinogenesis*. 2010 Mar;31(3):462-5.
IF (2010): 5.402
6. Falvella FS, Galvan A, Colombo F, **Frullanti E**, Pastorino U, Dragani TA. *Promoter Polymorphisms and Transcript Levels of Nicotinic Receptor CHRNA5*. *J Natl Cancer Inst*. 2010 Sep 8;102(17):1366-70.
IF (2010): 14.697
7. **Frullanti E**, Berking C, Harbeck N, Jézéquel P, Haugen A, Mawrin C, Parise OJr, Sasaki H, Tsuchiya N, Dragani TA. *Meta and pooled analyses of FGFR4 Gly388Arg polymorphism as a cancer prognostic factor*. *Eur J Cancer Prev*. 2011 Jul; 20(4):340-7.
IF (2011): 2.130
8. **Frullanti E**, Galvan A, Falvella FS, Manenti G, Colombo F, Vannelli A, Incarbone M, Alloisio M, Nosotti M, Santambrogio L, Gonzalez-Neira A, Pastorino U, Dragani TA. *Multiple genetic loci modulate lung adenocarcinoma clinical staging*. *Clin Cancer Res*. 2011 Apr 15;17(8):2410-6.
IF (2011): 7.742
9. Colombo F, Falvella FS, Galvan A, **Frullanti E**, Kunitoh H, Ushijima T, Dragani TA. *A 5'-region polymorphism modulates promoter activity of the tumor suppressor gene MFSD2A*. *Mol Cancer*. 2011 Jul 7;10(1):81.
IF (2011): 3.993
10. **Frullanti E**, Colombo F, Falvella FS, Galvan A, De Cecco L, Noci S, Incarbone M, Alloisio M, Tosi D, Nosotti M, Santambrogio, Pastorino U, Dragani TA. *Association of lung adenocarcinoma clinical stage with gene expression pattern in non-involved lung tissue*. *Int J Cancer*. 2012 Sep 1;131(5):E643-8.
IF (2011): 5.444
11. **Frullanti E**, La Vecchia C, Dragani TA, Boffetta P, Zocchetti C. *Vinyl chloride exposure and cirrhosis: a meta-analysis*. *Dig Liver Dis*. 2012 Sep;44(9):775-9.
IF (2011): 3.054
12. **Frullanti E**, La Vecchia C, Dragani TA, Boffetta P, Zocchetti C. *Authors' reply: Comment to "Vinyl chloride exposure and cirrhosis: a systematic review and meta-analysis"*. *Dig Liver Dis*. 2013 Aug;45(8):702
IF (2012): 3.054
13. Galvan A, **Frullanti E** [contributed equally to the work], Anderlini M, Manenti G, Noci S, Dugo M, Ambrogio F, De Cecco L, Spinelli R, Piazza R, Pirola A, Gambacorti-Passerini C, Incarbone M, Alloisio M, Santambrogio L, Nosotti M, Tosi T, Pastorino U, Dragani TA. *Gene expression signature of normal lung tissue associated with survival in lung adenocarcinoma patients*. *Carcinogenesis*. 2013 Dec;34(12):2767-73.
IF (2012):5.702
14. Mirella Filocamo, Chiara Baldo, Stefano Goldwurm, Alessandra Renieri, Corrado Angelini, Maurizio Moggio, Marina Mora, Giuseppe Merla, Luisa Politano, Barbara Garavaglia, Lorena Casareto, Franca Dagna Bricarelli, Fabio Corsoloni, Sara Galotto, Raffaella Mazzotti, Giorgia Stroppiana, Mauro Castagnetta, Massimo Moggi, Valeria Viotti, Alba Bonetti, Franca Felici, Francesca Natuzzi, Sonia Amabile, Elisa **Frullanti**, Iliara Meloni, Marina Fanin, Annachiara Nascimbeni, Elena Pegoraro, Enrico Peterle, Laura Napoli, Michela Ripolone, Monica Sciacco, Raffaella Violano, Eleonora Canioni, Sara Gibertini, Simona Saredi, Simona Zanotti, Carmela Fusco, Lucia Micale, Maria Teresa Pellico, Leopoldo Zelante, Paola D'Ambrosio, Esther Picillo, Antonella Taglia, Chiara Barzaghi, Celeste Panteghini¹⁰, Lorella Valletta for Telethon Network of Genetic Biobanks Staff. *Telethon Network of Genetic Biobanks: a Key Service for Diagnosis and Research on Rare Diseases*. *Orphanet J Rare Dis*. 2013 Aug 30;8(1):129.
IF (2012): 5.074
15. Disciglio V, Lo Rizzo C, Mencarelli MA, Mucciolo M, Marozza A, Di Marco C, Massarelli A, Canocchi V, Baldassari M, Ndoni E, **Frullanti E**, Amabile S, Anderlid BM, Metcalfe K, Le Caignec C, David A, Fryer A, Boute O, Joris A, Greco D, Pecile V, Battini R, Novelli A, Fichera M, Romano C, Mari F, Renieri A. *Interstitial 22q13 Deletions not involving SHANK3 gene: A new contiguous gene syndrome*. *Am J Med Genet A*. 2014 Jul;164A(7):1666-76.
IF (2012): 2.391

16. Dassano A, Colombo F, Trincucci G, **Frullanti E**, Galvan A, Pettinicchio A, De Cecco L, Borrego A, Martinez Ibanez O, Dragani TA, Manenti G. *Mouse Pulmonary Adenoma Susceptibility 1 Locus is an Expression QTL Modulating Kras-4A*. PLoS Genet. 2014 Apr 17;10(4):e1004307.
IF (2012): 8.517
17. Renieri A, Mencarelli MA, Cetta F, Baldassarri M, Mari F, Furini S, Piu P, Ariani F, Dragani TA, **Frullanti E**. *Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients*. Lung Cancer. 2014 Aug;85(2):168-74.
IF (JCR 2012): 3.434
18. Galvan A, Colombo F, **Frullanti E**, Dassano A, Noci S, Wang Y, Eisen T, Matakidou A, Tomasello L, Vezzalini M, Sorio C, Dugo M, Ambrogi F, Iacobucci I, Martinelli G, Incarbone M, Alloisio M, Nosotti M, Tosi D, Santambrogio L, Pelosi G, Pastorino U, Houlston RS, Dragani TA. *Germline polymorphisms and survival of lung adenocarcinoma patients: a genome-wide study in two European patient series*. Int J Cancer. 2015 Mar 1;136(5):E262-71.
IF (JCR 2012): 6.198
19. **Frullanti E**, Amabile S, Lolli MG, Bartolini A, Livide G, Landucci E, Mari F, Vaccarino FM, Ariani F, Massimino L, Renieri A, Meloni I. *Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice*. Eur J Hum Genet. 2016 Feb;24(2):252-7
IF (JCR 2012): 4.4
20. Patriarchi T, Amabile S, **Frullanti E**, Landucci E, Lo Rizzo C, Ariani F, Costa M, Olimpico F, W Hell J, M Vaccarino F, Renieri A, Meloni I. *Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1+/- patients and in foxg1+/- mice*. Eur J Hum Genet. 2016 Jun;24(6):871-80.
IF (JCR 2012): 4.4
21. Imperatore V, Mencarelli MA, Fallerini C, Bianciardi L, Ariani F, Furini S, Renieri A, Mari F, **Frullanti E**. *Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress*. Int J Mol Sci. 2016 Feb 27;17(3).
IF (JCR 2012): 2.464
22. Bianciardi L, Imperatore V, Fernandez-Vizarrá E, Lopomo A, Falabella M, Furini S, Galluzzi P, Grosso S, Zeviani M, Renieri A, Mari F, **Frullanti E**. *Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene*. Mol Genet Metab. 2016 Nov;119(3):214-222.
IF (JCR 2014): 2.625
23. Mucciolo M, Di Marco C, Canitano R, Buoni S, Frullanti E, Mencarelli MA, Bizzarri V, Amabile S, Radice L, Baldassarri M, Lo Rizzo C, Meloni I, Hayek J, Renieri A, Mari F. *A Genome Wide Copy Number Variations Analysis in Autism Spectrum Disorder (Asd) and Intellectual Disability (Id) in Italian Families*. J Genet Syndr Gene Ther. 2016, 7:5.
IF (JCR 2014): 2.05
24. Fallerini C, Baldassarri M, Trevisson E, Morbidoni V, La Manna A, Lazzarin R, Pasini A, Barbano G, Pinciaroli AR, Garosi G, **Frullanti E**, Pinto AM, Mencarelli MA, Mari F, Renieri A, Ariani F. *Alport syndrome: impact of digenic inheritance in patients management*. Clin Genet. 2017 Jul;92(1):34-44.
IF (JCR 2014): 3.931
25. Baldassarri M, Fallerini C, Cetta F, Ghisalberti M, Bellan C, Furini S, Spiga O, Crispino S, Gotti G, Ariani F, Paladini P, Renieri A, **Frullanti E**. *"Omic" Approach in Non-Smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine*. Cancer Res Treat. 2018 Apr;50(2):356-365.
IF (JCR 2016): 3.772
26. Cetta F, Renieri A, **Frullanti E**. *Germline mutations in lung cancer and personalized medicine*. Fam Cancer. 2018 Jul;17(3):429-430.
IF (JCR 2016): 1.66
27. Daga S, Baldassarri M, Lo Rizzo C, Fallerini C, Imperatore V, Longo I, **Frullanti E**, Landucci E, Massella L, Pecoraro C, Garosi G, Ariani F, Mencarelli MA, Mari F, Renieri A, Pinto AM. *Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome*. Hum Mutat. 2018 Feb;39(2):302-314.

IF (JCR 2016): 4.601

28. Papa FT, Mancardi MM, **Frullanti E**, Fallerini C, Della Chiara V, Zalba-Jadraque L, Baldassarri M, Gamucci A, Mari F, Veneselli E, Renieri A. *Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy*. Clin Dysmorphol. 2018 Jan;27(1):18-20.
IF (JCR 2016): 0.573
29. Pinto AM, Papa FT, **Frullanti E**, Meloni I, Tita R, Caselli R, Fallerini C, Lopergolo D, Cetta F, Mencarelli MA, Bocchia M, Gozzetti A, Renieri A. *Low-level TP53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia*. Br J Haematol. 2018 Feb 20. doi: 10.1111/bjh.15147.
IF (JCR 2016): 5.67
30. Imperatore V, Pinto AM, Gelli E, Trevisson E, Morbidoni V, **Frullanti E**, Hadjistilianou T, De Francesco S, Toti P, Gusson E, Roversi G, Accogli A, Capra V, Mencarelli MA, Renieri A, Ariani F. *Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma*. Eur J Hum Genet. 2018 Jul;26(7):1026-1037.
IF (JCR 2016): 4.287
31. Landucci E, Brindisi M, Bianciardi L, Catania LM, Daga S, Croci S, **Frullanti E**, Fallerini C, Butini S, Brogi S, Furini S, Melani R, Molinaro A, Lorenzetti FC, Imperatore V, Amabile S, Mariani J, Mari F, Ariani F, Pizzorusso T, Pinto AM, Vaccarino FM, Renieri A, Campiani G, Meloni I. *iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated α -tubulin defect which improves after iHDAC6 treatment in Rett syndrome*. Exp Cell Res. 2018 Jul 15;368(2):225-235.
IF (JCR 2016): 3.546
32. Cetta F, Palmieri M, Renieri A, **Frullanti E**. *Commentary: Potential Links between Hepadnavirus and Bornavirus Sequences in the Host Genome and Cancer*. Front Microbiol. 2018 Jul 23; doi.org/10.3389/fmicb.2018.01649
IF (JCR 2016): 4.076
33. Giliberti A, Currò A, Papa FT, **Frullanti E**, Ariani F, Coriolani G, Grosso S, Renieri A, Mari F. *MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype*. Eur J Med Genet. 2019 Feb 5. pii: S1769-7212(18)30600-1.
IF (JCR 2016): 2.137
34. Gelli E, Colombo M, Pinto AM, De Vecchi G, Foglia C, Amitrano S, Morbidoni V, Imperatore V, Manoukian S, Baldassarri M, Lo Rizzo C, Catania L, **Frullanti E**, Tagliafico E, Cortesi L, Spaggiari F, Mencarelli MA, Trevisson E, Radice P, Renieri A, Ariani F. *Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of BRCA1/2 Variants of Uncertain Significance*. Cancers (Basel). 2019 Mar 1;11(3). pii: E295.
IF (JCR 2016): 5.326
35. **Frullanti E**, Papa FT, Grillo E, Clarke A, Ben-Zeev B, Pineda M, Bahi-Buisson N, Bienvenu T, Armstrong J, Roche Martinez A, Mari F, Nissenkorn A, Lo Rizzo C, Veneselli E, Russo S, Vignoli A, Pini G, Djuric M, Bisgaard AM, Ravn K, Bosnjak VM, Hayek J, Khajuria R, Montomoli B, Cogliati F, Pintaudi M, Hadzsiev K, Craiu D, Voinova V, Djukic A, Villard L, Renieri A. *Analysis of the Phenotypes in the Rett Networked Database*. Int J Genomics. 2019 Mar 27;2019:6956934. doi: 10.1155/2019/6956934.
IF (JCR 2016): 2.402
36. Palmieri M, Baldassarri M, Fava F, Fabbiani A, Campennì GM, Mencarelli MA, Tita R, Marsili S, Renieri A, **Frullanti E**. *PIK3CA-CDKN2A clonal evolution in metastatic breast cancer and multiple points cell-free DNA analysis*. Cancer Cell Int. 2019 Oct 28;19:274. doi: 10.1186/s12935-019-0991-y.
IF (JCR 2016): 2.740
37. Daga S, Donati F, Capitani K, Croci S, Tita R, Giliberti A, Valentino F, Benetti E, Fallerini C, Niccheri F, Baldassarri M, Mencarelli MA, **Frullanti E**, Furini S, Conticello SG, Renieri A, Pinto AM. *New frontiers to cure Alport syndrome: COL4A3 and COL4A5 gene editing in podocyte-lineage cells*. Eur J Hum Genet. 2019 Nov 21. doi: 10.1038/s41431-019-0537-8. [Epub ahead of print]
IF (JCR 2016): 4.287
38. Pinto AM, Daga S, Fallerini C, Bruttini M, Baldassarri M, Giliberti A, **Frullanti E**, Guarnieri A, Garosi G,

Renieri A. Detection of Cryptic Mosaicism in X-linked Alport Syndrome Prompts to Reevaluate Living-donor Kidney Transplantation. Transplantation. 2019 Dec 31. doi: 10.1097/TP.0000000000003104.
IF (JCR 2018): 3.960

39. Palmieri M, Baldassarri M, Fava F, Fabbiani A, Gelli E, Tita R, Torre P, Petrioli R, Hadjistilianou T, Galimberti D, Cinotti E, Bengala C, Mandalà M, Piu P, Miano ST, Martellucci I, Vannini A, Pinto AM, Mencarelli MA, Marsili S, Renieri A, **Frullanti E**. Two-point-NGS analysis of cancer genes in cell-free DNA of metastatic cancer patients. Cancer Med. 2020 Jan 28. doi: 10.1002/cam4.2782.
IF (JCR 2018): 3.202

Total Impact Factor = 179.019

Impact Factor Mean = 4.59

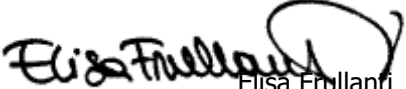
Number of Total Citation = 379

H-index (update 09 Dec 2019) = 11

G-index (update 09 Dec 2019) = 20

The undersigned, aware of the penalties provided for by the penal code, and by special laws against anyone who issues false declarations, also aware of the possibility of losing the benefits resulting from any provisions issued on the basis of untruthful declaration, declares that everything stated in the curriculum vitae corresponds to truth - art. 46, D.P.R. 445/2000.

Siena, 28/02/2020


Elisa Frullanti