

## Europass Curriculum Vitae



### Personal information

First name and Surname **Raffaella Cascella**

E-mail r.cascella@unizkm.al

Nationality Italian

Date of birth 02/02/1984

Gender Female

**Desired employment / Occupational field** Research Activity and Diagnostic Activity

### Work experience

Occupation or position held Associate Professor at Catholic University Our Lady of Good Counsel, Tirana, Albania.

Winner of the "Young Researchers" award at the University of Rome "Tor Vergata"

Main activities and responsibilities Medical Genetics, Genomic and Personalized Medicine, Ocular Genetics, Pharmacogenetics

Name and address of employer Catholic University Our Lady of Good Counsel, Laprakë, Rruga Dritan Hoxha, 1000, Tirana, Albania.

### Education and training

Dates 10/10/2010 - 10/10/2013

Title of qualification awarded PhD Student

Principal subjects/occupational skills covered Medical Genetics

Name and type of organisation providing education and training "Tor Vergata" University

Dates and Title of qualification awarded 14/05/2014 Professional Register of the National Order of Biologists, Section A, n. registration AA\_070959

01/10/2007 - 11/12/2009 2<sup>nd</sup> level degree in Genomic Biotechnology (LS, class 8/S), with 110 cum laude / 110

10/10/2003 - 10/07/2007 Bachelor's Degree in Industrial Biotechnology with 110 cum laude / 110

**Personal skills and competences**

Mother tongue(s) **Italian**

Other language(s) **English**

Self-assessment  
*European level (\*)*

**Language**

Understanding		Speaking		Writing
Listening	Reading	Spoken interaction	Spoken production	
B1	B2	B1	B2	B2

(\*) [Common European Framework of Reference for Languages](#)

Technical skills and competences

Excellent knowledge of molecular methods used in Medical Genetics laboratories:

- DNA extraction (manual and automated methods),
- RNA extraction (manual and automated methods),
- DNA amplification (PCR), Real Time PCR (allelic discrimination, quantification and expression),
- DNA separation methods (gel electrophoresis, capillary electrophoresis),
- DNA purification methods (chemical and enzymatic),
- Automated sequencing and new generation sequencing
- Analysis of microsatellites
- Bivariate and multivariate statistical analysis
- Bioinformatics analysis
- Management of Biobanks
- Excellent ability to develop molecular assays applicable in the diagnostic field (UPD15 Multiplex Assay, UPD7 Multiplex Assay, 26Cx, 26-30Cx, NAFLD, IL28B, HLA-B \* 57: 01)

Organisational skills and competences

Excellent knowledge of Mendelian hereditary diseases and multifactorial diseases. Study of chronic inflammatory diseases such as Atopic Eczema, Psoriasis, Arthropathic Psoriasis and Age-related Macular Degeneration. Study of neurodegenerative and neuromuscular diseases (FSHD). Remarkable experience in Ocular Genetics with the study of Mendelian and complex diseases (Pattern Dystrophy, Best Disease, Stargardt's Disease, Retinitis Pigmentosa, Keratoconus, Glaucoma and Age-Related Macular Degeneration).

Experience in the care activity and in the management of patients with different genetic pathologies.

Computer skills and competences

Excellent knowledge of the main operating software and the most important statistical analysis and data bioinformatics programs.

Other skills and competences

Excellent communication skills.  
Excellent decision-making and self-management skills.

Driving licence

Driver's license

## Bibliographic Parameters

### Scopus:

Total number of international publications: 66

h-index: 15

Total citations: 754

## Annexes

### Scientific publications

Longitudinal Structure-Function Evaluation in a Patient with CDHR1-Associated Retinal Dystrophy: Progressive Visual Function Loss with Retinal Remodeling. Cusumano A, Falsini B, D'Apolito F, D'Ambrosio M, Sebastiani J, Cascella R, Barati S, Giardina E. *Diagnostics (Basel)*. 2023;13(3):392. doi: 10.3390/diagnostics13030392.

Analysis of Genetic Variants Associated with COVID-19 Outcome Highlights Different Distributions among Populations. Fabrizio C, Termine A, Caputo V, Megalizzi D, Calvino G, Trastulli G, Ingrassi A, Ferrante S, Peconi C, Rossini A, Salvia A, Caltagirone C, Strafella C, Giardina E, Cascella R. *J Pers Med*. 2022;12(11):1851. doi: 10.3390/jpm12111851

D4Z4 Methylation Levels Combined with a Machine Learning Pipeline Highlight Single CpG Sites as Discriminating Biomarkers for FSHD Patients. *Cells*. Caputo V, Megalizzi D, Fabrizio C, Termine A, Colantoni L, Bax C, Gimenez J, Monforte M, Tasca G, Ricci E, Caltagirone C, Giardina E, Cascella R, Strafella C. 2022;11(24):4114. doi: 10.3390/cells11244114.

Update on the Molecular Aspects and Methods Underlying the Complex Architecture of FSHD. Caputo V, Megalizzi D, Fabrizio C, Termine A, Colantoni L, Caltagirone C, Giardina E, Cascella R, Strafella C. *Cells*. 2022;11(17):2687. doi: 10.3390/cells11172687.

Relationship between Nutrition, Lifestyle, and Neurodegenerative Disease: Lessons from ADH1B, CYP1A2 and MTHFR. Barati S, Fabrizio C, Strafella C, Cascella R, Caputo V, Megalizzi D, Peconi C, Mela J, Colantoni L, Caltagirone C, Termine A, Giardina E. *Genes (Basel)*. 2022;13(8):1498. doi:10.3390/genes13081498.

WARE: Wet AMD Risk-Evaluation Tool as a Clinical Decision-Support System Integrating Genetic and Non-Genetic Factors. Fabrizio C, Termine A, Caputo V, Megalizzi D, Zampatti S, Falsini B, Cusumano A, Eandi CM, Ricci F, Giardina E, Strafella C, Cascella R. *J Pers Med*. 2022;12(7):1034. doi: 10.3390/jpm12071034.

A Hybrid Machine Learning and Network Analysis Approach Reveals Two Parkinson's Disease Subtypes from 115 RNA-Seq Post-Mortem Brain Samples. Termine A, Fabrizio C, Strafella C, Caputo V, Petrosini L, Caltagirone C, Cascella R, Giardina E. *Int J Mol Sci*. 2022;23(5):2557. doi: 10.3390/ijms23052557.

Identification of Genetic Networks Reveals Complex Associations and Risk Trajectory Linking Mild Cognitive Impairment to Alzheimer's Disease. Strafella C, Caputo V, Termine A, Fabrizio C, Calvino G, Megalizzi D, Ruffo P, Toppi E, Banaj N, Bassi A, Bossù P, Caltagirone C, Spalletta G, Giardina E, Cascella R. *Front Aging Neurosci*. 2022;14:821789. doi: 10.3389/fnagi.2022.821789.

Tracking the Initial Diffusion of SARS-CoV-2 Omicron Variant in Italy by RTPCR and Comparison with Alpha and Delta Variants Spreading. Caputo V, Calvino G, Strafella C, Termine A, Fabrizio C, Trastulli G, Ingrassi A, Peconi C, Bardini S, Rossini A, Salvia A, Borsellino G, Battistini L, Caltagirone C, Cascella R, Giardina E. *Diagnostics (Basel)*. 2022;12(2):467. doi: 10.3390/diagnostics12020467.

Deregulation of ncRNA in Neurodegenerative Disease: Focus on circRNA, lncRNA and miRNA in Amyotrophic Lateral Sclerosis. Ruffo P, Strafella C, Cascella R, Caputo V,

Conforti FL, Andò S, Giardina E. *Front Genet.* 2021. doi: 10.3389/fgene.2021.784996. eCollection 2021.

Epigenomic signatures in age-related macular degeneration: Focus on their role as disease modifiers and therapeutic targets. Caputo V, Strafella C, Termine A, Fabrizio C, Ruffo P, Cusumano A, Giardina E, Ricci F, Cascella R. *Eur J Ophthalmol.* 2021. doi: 10.1177/11206721211028054.

Age and Sex Modulate SARS-CoV-2 Viral Load Kinetics: A Longitudinal Analysis of 1735 Subjects. Caputo V, Termine A, Fabrizio C, Calvino G, Luzzi L, Fusco C, Ingrassi A, Peconi C, D'Alessio R, Mihali S, Trastulli G, Megalizzi D, Cascella R, Rossini A, Salvia A, Strafella C, Giardina E. *J Pers Med.* 2021. doi: 10.3390/jpm11090882.

Precision Medicine into Clinical Practice: A Web-Based Tool Enables RealTime Pharmacogenetic Assessment of Tailored Treatments in Psychiatric Disorders. Zampatti S, Fabrizio C, Ragazzo M, Campoli G, Caputo V, Strafella C, Pellicano C, Cascella R, Spalletta G, Petrosini L, Caltagirone C, Termine A, Giardina E. *J Pers Med.* 2021. doi: 10.3390/jpm11090851.

Pharmacogenomics: An Update on Biologics and Small-Molecule Drugs in the Treatment of Psoriasis. Caputo V, Strafella C, Cosio T, Lanna C, Campione E, Novelli G, Giardina E, Cascella R. *Genes (Basel).* 2021. doi: 10.3390/genes12091398.

Case Report: Sars-CoV-2 Infection in a Vaccinated Individual: Evaluation of the Immunological Profile and Virus Transmission Risk. Strafella C, Caputo V, Guerrera G, Termine A, Fabrizio C, Cascella R, Picozza M, Caltagirone C, Rossini A, Balice MP, Salvia A, Battistini L, Borsellino G, Giardina E. *Front Immunol.* 2021. doi: 10.3389/fimmu.2021.708820.

Immune System and Neuroinflammation in Idiopathic Parkinson's Disease: Association Analysis of Genetic Variants and miRNAs Interactions. Strafella C, Caputo V, Termine A, Assogna F, Pellicano C, Pontieri FE, Macchiusi L, Minozzi G, Gambardella S, Centonze D, Bossù P, Spalletta G, Caltagirone C, Giardina E, Cascella R. *Front Genet.* 2021. doi: 10.3389/fgene.2021.651971.

Genetic Determinants Highlight the Existence of Shared Etiopathogenetic Mechanisms Characterizing Age-Related Macular Degeneration and Neurodegenerative Disorders. Strafella C, Caputo V, Termine A, Fabrizio C, Ruffo P, Potenza S, Cusumano A, Ricci F, Caltagirone C, Giardina E, Cascella R. *Front Neurol.* 2021. doi: 10.3389/fneur.2021.626066.

Genetic Counselling Improves the Molecular Characterisation of Dementing Disorders. Zampatti S, Ragazzo M, Peconi C, Luciano S, Gambardella S, Caputo V, Strafella C, Cascella R, Caltagirone C, Giardina E. *J Pers Med.* 2021.

Multi-Layer Picture of Neurodegenerative Diseases: Lessons from the Use of Big Data through Artificial Intelligence. Termine A, Fabrizio C, Strafella C, Caputo V, Petrosini L, Caltagirone C, Giardina E, Cascella R. *J Pers Med.* 2021. doi: 10.3390/jpm11040280.

Comparative analysis of antigen and molecular tests for the detection of SarsCoV-2 and related variants: A study on 4266 samples. Caputo V, Bax C, Colantoni L, Peconi C, Termine A, Fabrizio C, Calvino G, Luzzi L, Panunzi GG, Fusco C, Strafella C, Cascella R, Battistini L, Caltagirone C, Salvia A, Sancesario G, Giardina E. *Int J Infect Dis.* 2021. doi: 10.1016/j.ijid.2021.04.048.

Genetic Variants Allegedly Linked to Antisocial Behaviour Are Equally Distributed Across Different Populations. Zampatti S, Ragazzo M, Fabrizio C, Termine A, Campoli G, Caputo

V, Strafella C, Cascella R, Caltagirone C, Giardina E. *J Pers Med*. 2021. doi: 10.3390/jpm11030213.

Characterization of a natural variant of human NDP52 and its functional consequences on mitophagy. Di Rita A, Angelini DF, Maiorino T, Caputo V, Cascella R, Kumar M, Tiberti M, Lambrughi M, Wesch N, Löhr F, Dötsch V, Carinci M, D'Acunzo P, Chiurchiù V, Papaleo E, Rogov VV, Giardina E, Battistini L, Strappazon F. *Cell Death Differ*. 2021. doi: 10.1038/s41418-021-00766-3.

Investigation of Genetic Variations of IL6 and IL6R as Potential Prognostic and Pharmacogenetics Biomarkers: Implications for COVID-19 and Neuroinflammatory Disorders. Strafella C, Caputo V, Termine A, Barati S, Caltagirone C, Giardina E, Cascella R. *Life (Basel)*. 2020. doi: 10.3390/life10120351.

Overview of the molecular determinants contributing to the expression of Psoriasis and Psoriatic Arthritis phenotypes. Caputo V, Strafella C, Termine A, Dattola A, Mazzilli S, Lanna C, Cosio T, Campione E, Novelli G, Giardina E, Cascella R. *J Cell Mol Med*. 2020. doi: 10.1111/jcmm.15742.

Analysis of ACE2 Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. Strafella C, Caputo V, Termine A, Barati S, Gambardella S, Borgiani P, Caltagirone C, Novelli G, Giardina E, Cascella R. *Genes (Basel)*. 2020;11(7):741. doi: 10.3390/genes11070741.

Laryngopharyngeal reflux disease in adult patients: tears and pepsin. Magliulo G, Pace A, Plateroti R, Plateroti AM, Cascella R, Solito C, Rossetti V, Iannella G. *J Biol Regul Homeost Agents*. 2020;34(2):715-720. doi: 10.23812/19-437-L-26.

Shared (epi)genomic background connecting neurodegenerative diseases and type 2 diabetes. Caputo V, Termine A, Strafella C, Giardina E, Cascella R. *World J Diabetes*. 2020;11(5):155-164. doi: 10.4239/wjd.v11.i5.155.

Genetic Counseling and NGS Screening for Recessive LGMD2A Families. Strafella C, Caputo V, Campoli G, Galota RM, Mela J, Zampatti S, Minozzi G, Sancricca C, Servidei S, Giardina E, Cascella R. *High Throughput*. 2020;9(2):13. doi: 10.3390/ht9020013.

RNAseq-Based Prioritization Revealed COL6A5, COL8A1, COL10A1 and MIR146A as Common and Differential Susceptibility Biomarkers for Psoriasis and Psoriatic Arthritis: Confirmation from Genotyping Analysis of 1417 Italian Subjects. Caputo V, Strafella C, Termine A, Campione E, Bianchi L, Novelli G, Giardina E, Cascella R. *Int J Mol Sci*. 2020;21(8):2740. doi: 10.3390/ijms21082740.

Defective proteasome biogenesis into skin fibroblasts isolated from Rett syndrome subjects with MeCP2 non-sense mutations. Sbardella D, Tundo GR, Cunsolo V, Grasso G, Cascella R, Caputo V, Santoro AM, Milardi D, Pecorelli A, Ciaccio C, Di Pierro D, Leoncini S, Campagnolo L, Pironi V, Oddone F, Manni P, Foti S, Giardina E, De Felice C, Hayek J, Curatolo P, Galasso C, Valacchi G, Coletta M, Graziani G, Marini S. *Biochim Biophys Acta Mol Basis Dis*. 2020;1866(7):165793. doi: 10.1016/j.bbadis.2020.165793.

The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. Strafella C, Caputo V, Galota RM, Campoli G, Bax C, Colantoni L, Minozzi G, Orsini C, Politano L, Tasca G, Novelli G, Ricci E, Giardina E, Cascella R. *Hum Mol Genet*. 2019 ;28(23):3912-3920. doi: 10.1093/hmg/ddz239.

Follicular occlusion tetrad in a male patient with pachyonychia congenita: clinical and genetic analysis. Musumeci ML, Fiorentini F, Bianchi L, Cascella R, Giardina E, Caputo V, Micali G. *J Eur Acad Dermatol Venereol*. 2019;33 Suppl 6:36-39. doi: 10.1111/jdv.15851.

Bilateral Retinal Angiomatous Proliferation in a Variant of Retinitis Pigmentosa. Aloe G, De Sanctis CM, Strafella C, Cascella R, Missiroli F, Cesareo M, Giardina E, Ricci F. *Case Rep Ophthalmol Med*. 2019;2019:8547962. doi: 10.1155/2019/8547962.

Atopic Eczema: Genetic Analysis of COL6A5, COL8A1, and COL10A1 in Mediterranean Populations. Strafella C, Caputo V, Minozzi G, Milano F, Arcangeli M, Sobhy N, Abdelmaksood R, Hashad D, Vakirlis E, Novelli G, Cascella R, Giardina E. *Biomed Res Int*. 2019 Jun 4;2019:3457898. doi: 10.1155/2019/3457898

Limb-Girdle Muscular Dystrophies (LGMDs): The Clinical Application of NGS Analysis, a Family Case Report. Strafella C, Campoli G, Galota RM, Caputo V, Pagliaroli G, Carboni S, Zampatti S, Peconi C, Mela J, Sancricca C, Primiano G, Minozzi G, Servidei S, Cascella R, Giardina E. *Front Neurol*. 2019 Jun 13;10:619. doi: 10.3389/fneur.2019.00619.

The Interplay between miRNA-Related Variants and Age-Related Macular Degeneration: EVIDENCE of Association of MIR146A and MIR27A. Strafella C, Errichiello V, Caputo V, Aloe G, Ricci F, Cusumano A, Novelli G, Giardina E, Cascella R. *Int J Mol Sci*. 2019 Mar 29;20(7). pii: E1578. doi: 10.3390/ijms20071578.

Doyme honeycomb retinal dystrophy - functional improvement following subthreshold nanopulse laser treatment: a case report. Cusumano A, Falsini B, Giardina E, Cascella R, Sebastiani J, Marshall J. *J Med Case Rep*. 2019 Jan 10;13(1):5. doi: 10.1186/s13256-018-1935-1.

Expression and potential role of cellular retinol binding protein I in psoriasis. Costanza G, Doldo E, Ferlosio A, Tarquini C, Passeri D, Cascella R, Bavetta M, Di Stefani A, Bonifati C, Agostinelli S, Centofanti F, Giardina E, Campione E, Bianchi L, Donati P, Morrone A, Orlandi A. *Oncotarget*. 2018. 4;9(95):36736-36749. doi: 10.18632/oncotarget.26314. IF: 4.78

Digenic Inheritance of Shortened Repeat Units of the D4Z4 Region and a Loss-of-Function Variant in SMCHD1 in a Family With FSHD. Cascella R, et al., *Front Neurol*. 2018; 9:1027. doi: 10.3389/fneur.2018.01027. IF: 3.5

Identification of Duchenne/Becker muscular dystrophy mosaic carriers through a combined DNA/RNA analysis. Zampatti S, Mela J, Peconi C, Pagliaroli G, Carboni S, Barrano G, Zito I, Cascella R, et al., *Prenat Diagn*. 2018;38(13):1096-1102. doi: 10.1002/pd.5369.

Application of Precision Medicine in Neurodegenerative Diseases. Strafella C, Caputo V, Galota MR, Zampatti S, Marella G, Mauriello S, Cascella R, Giardina E. *Front Neurol*. 2018; 9:701. doi: 10.3389/fneur.2018.00701.

Uncovering genetic and non-genetic biomarkers specific for exudative age-related macular degeneration: significant association of twelve variants. Cascella R, Strafella C, Longo G, Ragazzo M, Manzo L, De Felici C, Errichiello V, Caputo V, Viola F, Eandi CM, Staurenghi G, Cusumano A, Mauriello S, Marsella LT, Ciccacci C, Borgiani P, Sangiuolo F, Novelli G, Ricci F, Giardina E. *Oncotarget*, 2017.

KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Cascella R, Strafella C, Ragazzo M, Manzo L, Costanza G, Bowes J, Hüffmeier U, Potenza S, Sangiuolo F, Reis A, Barton A, Novelli G, Orlandi A, Giardina E. *Oncotarget*. 2017 8;8(56):95401-95411. doi: 10.18632/oncotarget.20727.

Towards the application of precision medicine in Age-Related Macular Degeneration. Cascella R, Strafella C, Caputo V, Errichiello V, Zampatti S, Milano F, Potenza S, Mauriello S, Novelli G, Ricci F, Cusumano A, Giardina E. *Prog Retin Eye Res*. 2017 29. pii: S1350-9462(17)30058-7. doi: 10.1016/j.

Assessing individual risk for AMD with genetic counseling, family history, and genetic testing. Cascella R, Strafella C, Longo G, Manzo L, Ragazzo M, De Felici C, Gambardella S, Marsella LT, Novelli G, Borgiani P, Sangiuolo F, Cusumano A, Ricci F, Giardina E. *Eye (Lond)*. 2018 ;32(2):446-450. doi: 10.1038/eye.2017.192.

Biomolecular index of therapeutic efficacy in psoriasis treated by anti-TNF alpha agents. Bianchi L, Costanza G, Campione E, Ruzzetti M, Di Stefani A, Diluvio L, Giardina E, Cascella R, Cordiali-Fei P, Bonifati C, Chiricozzi A, Novelli G, Ensoli F, Orlandi A. *G Ital Dermatol Venereol*. 2016.

Pharmacogenomics of multifactorial diseases: a focus on psoriatic arthritis. Cascella R, Strafella C, Longo G, Maccarone M, Borgiani P, Sangiuolo F, Novelli G, Giardina E. *Pharmacogenomics*. 2016 17(8):943-51. doi: 10.2217/pgs.16.20.

Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. Pietropoli A, Capogna MV, Cascella R, Germani C, Bruno V, Strafella C, Sarta S, Ticconi C, Marmo G, Gallaro S, Longo G, Marsella LT, Novelli A, Novelli G, Piccione E, Giardina E. *Hum Genomics*. 2016;10:9. doi: 10.1186/s40246-016-0066-2.

Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. Cascella R, Strafella C, Gambardella S, Longo G, Borgiani P, Sangiuolo F, Novelli G, Giardina E. *Electrophoresis*. 2015. doi: 10.1002/elps.201500346.

May some HCV genotype 1 patients still benefit from dual therapy? The role of very early HCV kinetics. Tontodonati M, Cento V, Polilli E, Colabattista C, Cascella R, et al., *New Microbiol*. 2015 Nov;38(4):491-7.

The Genetics and the Genomics of Primary Congenital Glaucoma. Cascella R, Strafella C, Germani C, Novelli G, Ricci F, Zampatti S, Giardina E. *Biomed Res Int*. 2015;2015:321291.

FLG (filaggrin) null mutations and sunlight exposure: Evidence of a correlation. Cascella R, Strafella C, Germani C, Manzo L, Marsella LT, Borgiani P, Sobhy N, Abdelmaksood R, Gerou S, Ioannides D, Sangiuolo F, Novelli G, Hashad D, Vakirlis E, Giardina E. *J Am Acad Dermatol*. 2015;73(3):528-9.

Comparative analysis between saliva and buccal swabs as source of DNA: lesson from HLA-B\*57:01 testing. Cascella R, Stocchi L, Strafella C, Mezzaroma I, Mannazzu M, Vullo V, Montella F, Parruti G, Borgiani P, Sangiuolo F, Novelli G, Pirazzoli A, Zampatti S, Giardina E. *Pharmacogenomics*. 2015;16(10):1039-46.

Absence of filaggrin mutation in a patient affected by pachyonychia congenita and mild atopic dermatitis. Terrinoni A, Giardina E, Pertusi G, Cascella R, Serra V, Bornacina C, Palombo R, Tiberio R, Gattoni M, Novelli G, Annicchiarico-Petruzzelli M, Melino G, Colombo E. *Eur J Dermatol*. 2014;24(6):703-4.

Age-related macular degeneration: insights into inflammatory genes. Cascella R, Ragazzo M, Strafella C, Missiroli F, Borgiani P, Angelucci F, Marsella LT, Cusumano A, Novelli G, Ricci F, Giardina E. *J Ophthalmol*. 2014;2014:582842.

Direct PCR: a new pharmacogenetic approach for the inexpensive testing of HLA-B\*57:01. Cascella R, Strafella C, Ragazzo M, Zampatti S, Borgiani P, Gambardella S, Pirazzoli A, Novelli G, Giardina E. *Pharmacogenomics J*. 2014. doi: 10.1038/tpj.2014.48.

Haplotypes in IL-8 Gene Are Associated to Age-Related Macular Degeneration: A Case-Control Study. Ricci F, Staurenghi G, Lepre T, Missiroli F, Zampatti S, Cascella R, Borgiani P, Marsella LT, Eandi CM, Cusumano A, Novelli G, Giardina E. *PLoS One*. 2013;8(6):e66978

Association Of Kif3a, But Not Ovol1 And Act19, With Atopic Eczema In Italian Patients. Lepre T, Cascella R, Ragazzo M, Galli E, Novelli G, Giardina E. *Br J Dermatol*. 2012. doi: 10.1111/bjd.12178.

The Pharmacogenomic HLA Biomarker Associated to Adverse Abacavir Reactions: Comparative Analysis of Different Genotyping Methods. Stocchi L, Cascella R, Zampatti S, Pirazzoli A, Novelli G, Giardina E. *Curr Genomics*. 2012;13(4):314-20. doi: 10.2174/138920212800793311.

Polymorphisms in ARMS2 (LOC387715) and LOXL1 genes in the Japanese with age-related macular degeneration. Lepre T, Cascella R, Missiroli F, De Felici C, Taglia F, Zampatti S, Cusumano A, Ricci F, Giardina E, Eandi CM, Novelli G. *Am J Ophthalmol*. 2011;152(2):325-6; author reply 326. doi: 10.1016/j.ajo.2011.04.021.

Full sequencing of the FLG gene in Italian patients with atopic eczema: evidence of new mutations, but lack of an association. Cascella R, Foti Cuzzola V, Lepre T, Galli E, Moschese V, Chini L, Mazzanti C, Fortugno P, Novelli G, Giardina E. *J Invest Dermatol*. 2011;131(4):982-4. doi: 10.1038/jid.2010.398.

A multiplex molecular assay for the detection of uniparental disomy for human chromosome 7.

Giardina E, Peconi C, Cascella R, Sinibaldi C, Foti Cuzzola V, Nardone AM, Bramanti P, Novelli G. *Electrophoresis*. 2009;30(11):2008-11. doi: 10.1002/elps.200800744.

A multiplex molecular assay for the detection of uniparental disomy for human chromosome 15. Giardina E, Peconi C, Cascella R, Sinibaldi C, Nardone AM, Novelli G. *Electrophoresis*. 2008;29(23):4775-9. doi: 10.1002/elps.200800047.

## Oral Communications

“Evidence of common and differential genetic biomarkers for Psoriasis and Psoriatic Arthritis” Congresso Mondiale di Dermatologia, Milano 2019.

La Donna ieri e oggi: il suo tempo, i suoi spazi. “La donna e la Genetica” Tirana 2019.

Chi dice donna dice.... Donna “Donna e Genetica”. Veroli 2018.

Generation Sequencing: applicazioni e stato dell’arte, “NGS e Malattie Oculari: test diagnostici e predittivi”. Pozzilli, 2017.

Aspetti Molecolari di Prevenzione e Salute, Istituto Superiore di Sanità, “Il contributo delle omiche alla medicina personalizzata”. Roma 2016.

Università degli studi di Roma “Tor Vergata”, Remembering Sergio Chimenti, *Genetics & Genomics of Psoriasis*. Roma, 2016.

International Congress of Dermatology dedicated to Psoriasis, PSOFUTURE, “Insights in psoriatic arthritis genetics: KIF3A as a new susceptibility locus”. Roma, 2015.

La formazione sanitaria tra etica e tecnologia, Il gene tra etica e diritto”. Roma, 2014.



**Teaching Activity**

Corso ECM 2014, Scuola Medica Ospedaliera “Potenzialità della Real Time PCR nel laboratorio di biologia molecolare”. Roma, 2014.

Professor of Medical Genetics, degree in Physiotherapy. Tirana

Professor of Medical Genetics, degree in nursing sciences. Tirana

Professor of Biology and Medical Genetics, degree in nursing sciences. Elbasan.

Lecturer, Master's Degree in Forensic Genetics, “Tor Vergata” University.

Level I Master Teacher in New Techniques and Strategies of Visual Rehabilitation, “Sapienza” University

Professor of Biology and Medical Genetics, Scuola Permanente in Biologia Forense, il direttore tecnico biologo nella Polizia di Stato: inquadramento normativo e definizione delle competenze.

Teaching activity and exercises at the Master of Science in Medical Biotechnology at the University of Rome "Tor Vergata".

Teaching activities and exercises Degree Course in Industrial Biotechnology at the University of Urbino "Carlo Bo".