Curriculum Vitae

Personal Data

Name: Raffaella

Surname: Cascella

Education

February 2022:

National scientific qualification for Associate Professor in the scientific disciplinary sector MED/03.

December 2021:

Specialization in Medical Genetics discussing the thesis entitled “Development and validation of two computational systems to be applied in clinical practice for the implementation of precision medicine protocols” at Medical Genetics Laboratory, University of Rome Tor Vergata.

June 2014:

PhD in Advanced Technologies in Biomedicine (XXVI cycle) discussing the thesis entitled “Genetic characterization of Age-related Macular Degeneration: clinical and molecular basis for the development of a predictive test” at Medical Genetics Laboratory, University of Rome Tor Vergata.

Registration in the Professional Register of the National Order of Biologists.

December 2009:

Master’s Degree in Genomic Biotechnology discussing the thesis entitled “Mutational analysis of the FLG gene in Italian patients with Atopic Eczema. Case-control study and genotype-phenotype correlation” at University of Rome “Sapienza”.

July 2007:

Bachelor’s degree in Industrial biotechnology addressed to the production of diagnostics, therapeutics and vaccines discussing the thesis entitled “Development of fast and inexpensive protocol for the detection of Uniparental Disomies in the pre- and postnatal era” at University of Urbino “Carlo Bo”

Position and Work Experience

September 2016 – February 2023:

Research grants on the topics “Research of genomic biomarkers in the field of ocular diseases; Research of genomic biomarkers in the field of neurodegenerative diseases; Research of genomic biomarkers and application of NSG protocols for the study of Klinefelter syndrome” at Medical Genetics Laboratory, University of Rome Tor Vergata.

March 2018 – March 2022:

Teaching assistant of Medical Genetics (MED/03). Master's degree in Medical Biotechnology, University of Rome Tor Vergata.

March 2018 – Present:

Teaching assistant of Medical Genetics (MED/03). Degree in Medicine and Surgery, University of Rome Tor Vergata.

June 2015 – June 2016:

Postgraduate scholarship on the topic "Genetic counseling in predictive ocular tests" at Medical Genetics Laboratory, University of Rome Tor Vergata.

November 2014 – November 2015:

Coordinated and continuous collaboration for research activity on "The genetics of hereditary retinal degeneration", at University of Rome "Sapienza".

Teaching Activity

May 2015 and May 2017

Lecturer of fundamentals of Molecular Biology and Human Genetics at the Permanent School in Forensic Biology. The biologist technical director in the State Police: regulatory framework and definition of skills.

January 2014 – Present:

Lecturer of Master's Degree in Forensic Genetics, University of Rome Tor Vergata. Topics: "Introduction to the Medical Genetics", "DNA Extraction Methods" and "Molecular Techniques".

June 2015 – Present:

Lecturer of Master in New Techniques and Strategies of Visual Rehabilitation, "Sapienza" University of Rome. Topic of the lesson "The genetics of eye diseases".

October 2018 – Present:

Professor of Medical Genetics, degree course in Physiotherapy at the Catholic University "Our Lady of Good Counsel" Tirana, Albania.

Professor of Medical Genetics, degree course in Nursing at the Catholic University "Our Lady of Good Counsel" Tirana, Albania.

Scientific activity

Research Activity:

Excellent skills in the development of molecular assays to apply in the clinical practice. Molecular assays developed: UPD15 Multiplex Assay, UPD7 Multiplex Assay, 26Cx, 26-30Cx, NAFLD, IL28B, HLA-B*57:01. Excellent knowledge of Mendelian inherited diseases and multifactorial diseases. In particular, the research activity is based on complex chronic-inflammatory diseases (Atopic Eczema, Psoriasis, Psoriatic Arthritis); familial and multifactorial ocular diseases (Pattern Dystrophy, Best's Disease, Stargardt's Disease, Retinitis Pigmentosa, Retinal Dystrophies, Keratoconus and Age-Related Macular Degeneration). Study of neuromuscular (FSHD, DMD, LGMD) and neurodegenerative (AD and PD) diseases. Excellent knowledge of the main operating software and of the most important biostatistics and bioinformatics analysis programs.

Scientific publication (2008 – 2023):

- 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.
- 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.
- 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.

- 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. doi: 10.1016/j.ajo.2011.04.021.
- 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.
- Association Of Kif3a, But Not Ovol1 And Actl9, 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.
- 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. doi: 10.1371/journal.pone.0066978.
- 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.
- 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. doi: 10.1155/2014/582842
- 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. doi: 10.1684/ejd.2014.2446.
- 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. doi: 10.2217/pgs.15.59.
- 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. doi: 10.1016/j.jaad.2015.06.022
- 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. doi: 10.1155/2015/321291.
- 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.
- 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.
- Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. Pietropolli 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.

- 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.
- 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. doi: 10.23736/S0392-0488.
- 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.
- 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.
- KIF3A and IL-4are 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.
- Uncovering genetic and non-genetic biomarkers specific for exudative age-related macular degeneration: significant association of twelve variants. Cascella R, Strafella C, Longo G, RagazzoM, 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;9(8):7812-7821. doi: 10.18632/oncotarget.23241.
- 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.
- 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, Marella G, Milano F, Arcangeli M, Caltagirone C, Novelli A, Giardina E. Prenat Diagn. 2018. doi: 10.1002/pd.5369.
- 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, Strafella C, Caputo V, Galota RM, Errichiello V, Scutifero M, Petillo R, Marella GL, Arcangeli M, Colantoni L, Zampatti S, Ricci E, Deidda G, Politano L, Giardina E. Front Neurol. 2018;9:1027. doi: 10.3389/fneur.2018.01027.
- 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;9(95):36736-36749. doi: 10.18632/oncotarget.26314.
- Doyne 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;13(1):5. doi: 10.1186/s13256-018-1935-1.
- 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;20(7). pii: E1578. doi: 10.3390/ijms20071578.

- 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;10:619. doi: 10.3389/fneur.2019.00619.
- 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; 2019:3457898. doi: 10.1155/2019/3457898.
- 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.
- 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.
- 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.
- 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.
- 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.
- 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.
- 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.
- 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.
- 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.
- 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;24(23):13554-13563. doi: 10.1111/jcmm.15742.

- 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;10(12):351. doi: 10.3390/life10120351.
- 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, Strappazzon F. Cell Death Differ. 2021;28(8):2499-2516. doi: 10.1038/s41418-021-00766-3.
- 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;11(3):213. doi: 10.3390/jpm11030213.
- 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;108:187-189. doi: 10.1016/j.ijid.2021.04.048.
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- 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;12:626066. doi: 10.3389/fneur.2021.626066.
- 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;12:651971. doi: 10.3389/fgene.2021.651971.
- 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;12:708820. doi: 10.3389/fimmu.2021.708820.
- 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;12(9):1398. doi: 10.3390/genes12091398.
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- 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, Ingrascì 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;11(9):882. doi: 10.3390/jpm11090882.

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- 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, Ingrascì 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.
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- 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.
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