Europass Curriculum Vitae

Personal information

First name(s) / Surname(s)	**Raffaella Cascella**
Address(es)	403, Tiburtina Valeria Street, 65128, Pescara, Italy
Telephone(s)	Mobile: +39 3381478346
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

Dates	2019 - 2017
Occupation or position held	Researcher at Catholic University Our Lady of Good Counsel, Tirana, Albania. Winner of the "Young Researchers" award at the University of Rome "Tor Vergata" Visiting lecturer on Medical Genetics (MED03), at 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 2nd 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**

Language	Understanding		Speaking		Writing
Self-assessment *European level (*)*	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: 33
h-index: 8
Total citations: 200

Annexes

Scientific publications

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.

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 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-ofFunction 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-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.

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. 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.

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 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.

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.

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

Teaching Activity

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".

Rome, 11/07/2019

Raffaella Cascella