

## ***CURRICULUM VITAE***

**NOME E COGNOME** CINZIA CICCACCI

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### **POSIZIONE ATTUALE**

Da Ottobre 2021 Professore Associato di Genetica Medica (Med/03) presso UniCamillus, Saint Camillus International University of Health Sciences

### **POSIZIONI PRECEDENTI**

- 2018-2021 Ricercatore RTDb in Genetica Medica presso UniCamillus, Saint Camillus International University of Health Sciences
- 2007-2018 Borsista e Assegnista di Ricerca presso Università di Tor Vergata (Dipartimento di Biomedicina e Prevenzione)
- 2006-2007 Assegnista di ricerca presso Filas spa
- 2005 Assegnista di Ricerca presso Università di Tor Vergata (Dipartimento di Biologia)
- 2001-2004 PhD presso Università di Tor Vergata (Dipartimento di Biologia)

### **STUDI EFFETTUATI ED ABILITAZIONI**

- Laurea in Scienze Biologiche con la votazione di 110 su 110 e lode conseguita il 08/03/2001 presso l'Università di Roma 'Tor Vergata'
- Esame di Stato per l'abilitazione e l'iscrizione all'Albo dei Biologi (giugno 2002)
- Dottorato di Ricerca in Biologia Evoluzionistica ed Ecologia (XVII ciclo) (novembre 2001-ottobre 2004)
- 26/05/2005 Conseguito il titolo di dottore di ricerca
- giugno 2010-giugno 2015 Scuola di specializzazione in Genetica Medica (indirizzo tecnico), Università degli studi di Roma "Tor Vergata", Facoltà di Medicina e Chirurgia
- 05/12/2014 Abilitazione scientifica nazionale per il settore 06/N1, II fascia
- 21-07-2015 Conseguito il titolo di specialista in Genetica Medica (indirizzo tecnico)
- 31/03/2017 Abilitazione scientifica nazionale per il settore 06/A1 (Genetica Medica), II fascia

### **ATTIVITA' DI RICERCA**

L'attività scientifica della dott.ssa Cinzia Ciccacci è documentata da 74 pubblicazioni su riviste internazionali "peer-reviewed", di cui 71 indicizzate su pubmed, per un IF totale di circa 300, citazioni totali = 1.414 (fonte Scopus) e con h-index=22 (fonte Scopus) e da 54 comunicazioni a Congressi Nazionali e Internazionali.

Tale attività si è focalizzata all'inizio della sua carriera accademica sulla genetica delle popolazioni umane e la genetica evoluzionistica con particolare riguardo a A) caratterizzazione del pattern di variabilità per SNS (Single Nucleotide Substitutions) MisSense e SameSense in sequenze codificanti; B)

variabilità alplotipica del gene CFTR (Cystic Fibrosis conductance Transmembrane Regulator) in varie popolazioni umane.

Negli ultimi 16 anni l'attività della dottoressa si è focalizzata sulla ricerca dei fattori genetici che contribuiscono A) alla suscettibilità alle malattie multifattoriali, (in particolare alle malattie infiammatorie croniche dell'intestino, al Lupus Sistemico Eritematoso, all'Artrite Reumatoide, al Diabete e alle sue complicanze) investigando il ruolo della variabilità genetica in relazione alla variabilità fenotipica di tali patologie e alle loro diverse manifestazioni cliniche e complicanze; B) alla variabilità nella risposta ai farmaci, sia in termini di efficacia che di tossicità (Farmacogenetica/Farmacogenomica), in particolare studiando i fattori genetici predittivi di risposta nei trattamenti con i farmaci Nevirapina (anti-retrovirale utilizzato nel trattamento dell'Hiv) e Warfarina (anticoagulante orale) e più recentemente con i farmaci biologici nelle malattie autoimmuni .

Negli ultimi 16 anni ha partecipato a diversi progetti di ricerca (riguardanti la genetica di patologie umane multifattoriali e la Farmacogenetica) finanziati dal MIUR e da altri Enti Nazionali e Internazionali.

**Reviewer** per numerose riviste tra cui: Plos One, Journal of Cardiovascular Medicine, Thrombosis Research, Pharmacogenomics, Journal of Immunology Research, International Journal of Environmental Research and Public Health, International Journal of Molecular Science, Pharmacogenomics Journal, Genetics Research International, Human Molecular Genetics.

**Reviewer** per progetti di Ricerca del “National Science Centre, Poland” e del “Health Research Council of New Zealand “

**Academic Editor per:**

Genes dal 2021

Journal of Immunology Research dal 2018

Plos One dal 2018

Guest Editor for “Pharmaceuticals” (2020-2022)

**Attività Didattica**

Dal 2018 ad oggi, Docente di Genetica Medica presso UniCamillus per i corsi di Laurea in Medicina, Ostetricia, Tecniche di Laboratorio Biomedico, Fisioterapia. Dal 2018 al 2020 docente di Genetica Medica anche per i corsi di Infermieristica e Tecniche di Radiologia Medica. Dal 2020 ad oggi Docente di Genetica Medica nel corso di Laurea in Odontoiatria. Coordinatore di diversi corsi integrati, membro di diverse commissioni di riconoscimento crediti e di trasferimento.

## ELENCO DELLE PUBBLICAZIONI

Modiano G, Bombieri C, Ciminelli BM, Belpinati F, Giorgi S, Georges MD, Scotet V, Pompei F, **Ciccacci C**, Guittard C, Audrezet MP, Begnini A, Toepfer M, Macek M, Ferec C, Claustres M, Pignatti PF. *A large-scale study of the random variability of a coding sequence: a study on the CFTR gene*. Eur J Hum Genet. 2005 Feb;13(2):184-92.

Pompei F, Ciminelli BM, Bombieri C, **Ciccacci C**, Koudova M, Giorgi S, Belpinati F, Begnini A, Cerny M, Des Georges M, Claustres M, Ferec C, Macek M, Modiano G, Pignatti PF. *Haplotype block structure study of the CFTR gene. Most variants are associated with the M470 allele in several European populations*. Eur J Hum Genet. 2006 Jan;14(1):85-93.

Ciminelli BM, Bonizzato A, Bombieri C, Pompei F, Gabaldo M, **Ciccacci C**, Begnini A, Holubova A, Zorzi P, Piskackova T, Macek M Jr, Castellani C, Modiano G, Pignatti PF. *Highly preferential association of NonF508del CF mutations with the M470 allele*. J Cyst Fibros. 2007 Jan;6(1):15-22.

Borgiani P, Perricone C, **Ciccacci C**, Romano S, Novelli G, Biancone L, Petruzzello C, Pallone F. *Interleukin-23R Arg381Gln is associated with susceptibility to Crohn's disease but not with phenotype in an Italian population*. Gastroenterology. 2007 Sep;133(3):1049-51.

Borgiani P, **Ciccacci C**, Forte V, Romano S, Federici G, Novelli G. *Allelic variants in the CYP2C9 and VKORC1 loci and interindividual variability in the anticoagulant dose effect of warfarin in Italians*. Pharmacogenomics. 2007 Nov;8(11):1545-1550.

Perricone C, Borgiani P, Romano S, **Ciccacci C**, Fusco G, Novelli G, Biancone L, Calabrese E, Pallone F. *ATG16L1 Ala197Thr is not associated with susceptibility to Crohn's disease or with phenotype in an Italian population*. Gastroenterology. 2008 Jan;134(1):368-70.

Novelli G, **Ciccacci C**, Borgiani P, Amati M P, Abadie E. Genetic tests and genomic biomarkers: regulation, qualification and validation. *Clinical Cases in Mineral and Bone Metabolism*. 2008; 5 (2): 149-154.

Borgiani P, **Ciccacci C**, Forte V, Sirianni E, Novelli L, Bramanti P, Novelli G. *CYP4F2 genetic variant (rs2108622) significantly contributes to warfarin dosing variability in the Italian population*. Pharmacogenomics. 2009; 10(2):261-6.

G. Novelli, P. Borgiani, C. **Ciccacci**, N. Di Daniele, G. Sirugo, M. Papaluca Amati. *Pharmacogenomics: role in medicines approval and clinical use*. Public Health Genomics 2010; 13(5):284-91

**Ciccacci C**, Borgiani P, Ceffa S, Sirianni E, Marazzi MC, Doro Altan AM, Paturzo G, Bramanti P, Novelli G, Palombi L. *Nevirapine-induced hepatotoxicity and Pharmacogenetics: a retrospective study in a population of Mozambique*. Pharmacogenomics. 2010 Jan;11(1):23-31

Predazzi IM, Martínez-Labarga C, Vecchione, L, Mango R, **Ciccacci C**, Amati F, Ottoni C, Crawford M, Rickards O, Romeo F, Novelli G. *Population differences in allele frequencies at the OLR1 locus may suggest geographic disparities in cardiovascular risk events*. Ann Hum Biol. 2010 Apr;37(2):136-48.

Ciminelli BM, Bombieri C, **Ciccacci C**, Belpinati F, Pompei F, Maselli R, Simporé J, Pignatti PF, Modiano G. *Anthropological features of the CFTR gene: Its variability in an African population*. Ann Hum Biol. 2011 Mar; 38(2):203-9.

**Ciccacci C**, Falconi M, Paolillo N, Oteri F, Forte V, Novelli G, Desideri A, Borgiani P. *Characterization of a novel CYP2C9 gene mutation and structural bioinformatic protein analysis in a warfarin hypersensitive patient*. Pharmacogenetics and Genomics. 2011, 21(6):344-346.

**C Ciccacci**, N Paolillo, D Di Fusco, G Novelli and P Borgiani. *EPHX1 Polymorphisms Are Not Associated with Warfarin Response in an Italian Population*. Clin Pharmacol Ther. 2011 Jun;89(6):791

**Ciccacci C**, Biancone L, Di Fusco D, Ranieri M, Condino G, Giardina E, Onali S, Lepre T, Pallone F, Novelli G, Borgiani P. *TRAF3IP2 gene is associated with cutaneous extraintestinal manifestations in Inflammatory Bowel Disease*. J Crohns Colitis. 2013; 7 (1):44-52.

**Ciccacci C**, Di Fusco D, Cacciotti L, Morganti R, D'Amato C, Novelli G, Sangiuolo F, Spallone V, Borgiani P. *TCF7L2 gene polymorphisms and type 2 diabetes: association with diabetic retinopathy and cardiovascular autonomic neuropathy*. Acta Diabetol. 2013; 50(5):789-99.

**Ciccacci C**, Di Fusco D, Marazzi MC, Liotta G, Palombi L, Novelli G, Borgiani P. *ABCC10 rs2125739 polymorphism and nevirapine-induced hepatotoxicity: lack of association in a population from Mozambique*. Pharmacogenet Genomics. 2013 Gen; 23(1):38-39

E Danese, M Montagnana, JA Johnson, AE Rettie, CF Zambon, SA Lubitz, G Suarez-Kurtz, LH Cavallari, L Zhao, M Huang, Y Nakamura, T Mushiroda, MK Kringen, P Borgiani, **C Ciccacci**, NT Au, T Langae, V Siguret, M-A Loriot, H Sagreiya, RB Altman, MHA Shahin, SA Scott, SI Khalifa, B Chowbay, IM Suriapranata, M Teichert, BH Stricker, M Taljaard, MR Botton, JE Zhang, M Pirmohamed, X Zhang, JF Carlquist, BD Horne, MTM Lee, V Pengo, GC Guidi, P Minuz and C Fava. *Impact of the CYP4F2 p. V433M Polymorphism on Coumarin Dose Requirement: Systematic Review and Meta-Analysis*. Clin Pharmacol Ther. 2012 Dec;92(6):746-56

**Ciccacci C**, Di Fusco D, Cacciotti L, Morganti R, D'Amato C, Greco C, Rufini S, Novelli G, Sangiuolo F, Spallone V, Borgiani P. *MicroRNA genetic variations: association with type 2 diabetes*. Acta Diabetol. 2013, 50 (6):867-872

Davide Di Fusco, **Cinzia Ciccacci**, Sara Rufini, Vittorio Forte, Giuseppe Novelli and Paola Borgiani. *Resequencing of VKORC1, CYP2C9 and CYP4F2 genes in Italian patients requiring extreme low and high warfarin doses*. Thromb Res. 2013 Jul;132(1):123-6.

**Cinzia Ciccacci**, Davide Di Fusco, Maria C. Marazzi, Ines Zimba, Fulvio Erba, Giuseppe Novelli, Leonardo Palombi, Paola Borgiani and Giuseppe Liotta. *Association between CYP2B6 polymorphisms and Nevirapine-induced SJS/TEN: a pharmacogenetics study*. Eur J Clin Pharmacol. 2013, 69 (11): 1909-1916.

Carlo Perricone, **Cinzia Ciccacci**, Fulvia Ceccarelli, Davide Di Fusco, Francesca Romana Spinelli, Enrica Cipriano, Giuseppe Novelli, Guido Valesini, Fabrizio Conti and Paola Borgiani. *TRAF3IP2 gene and systemic lupus erythematosus: association with disease susceptibility and pericarditis development*. Immunogenetics, 2013, 65(10):703-9.

- G Novelli, E Abadie, **C Ciccacci**, D Di Fusco, M Papaluca Amati, P Borgiani. *Biomarkers to predict drug efficacy and safety in neurodegenerative diseases*. Eur J Neurodegen Dis, 2013, 2 (1): 69-77
- Lucia Novelli, **Cinzia Ciccacci**, Paola Borgiani and Giuseppe Novelli. *Recent advances in the genetic susceptibility to Osteoarthritis*. Eur J Musculoskel Dis, 2013, 2 (1): 9-19
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- Livia Biancone, Emma Calabrese, Carmelina Petruzzello, Alessandra Capanna, Francesca Zorzi, Sara Onali, Giovanna Condino, Elisabetta Lolli, **Cinzia Ciccacci**, Paola Borgiani, Francesco Pallone. *A family study of asymptomatic small bowel Crohn's disease*. Dig Liver Dis. 2014 Mar;46(3):276-8.
- Ciccacci C**, Morganti R, Di Fusco D, D'Amato C, Cacciotti L, Greco C, Rufini S, Novelli G, Sangiuolo F, Marfia GA, Borgiani P, Spallone V. *Common polymorphisms in MIR146a, MIR128a and MIR27a genes contribute to neuropathy susceptibility in type 2 diabetes*. Acta Diabetol. 2014 Aug;51(4):663-71.
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- Ciccacci C**, Rufini S, Mancinelli S, Buonomo E, Giardina E, Scarcella P, Marazzi MC, Novelli G, Palombi L, Borgiani P. *A Pharmacogenetics Study in Mozambican Patients Treated with Nevirapine: Full Resequencing of TRAF3IP2 Gene Shows a Novel Association with SJS/TEN Susceptibility*. Int J Mol Sci. 2015 Mar 12;16(3):5830-8.
- Rufini S, **Ciccacci C**, Di Fusco D, Ruffa A, Pallone F, Novelli G, Biancone L, Borgiani P. *Autophagy and inflammatory bowel disease: Association between variants of the autophagy-related IRGM gene and susceptibility to Crohn's disease*. Dig Liver Dis. 2015 Sep;47(9):744-50.
- Ciccacci C**, Rufini S, Politi C, Novelli G, Forte V, Borgiani P. *Could MicroRNA polymorphisms influence warfarin dosing? A pharmacogenetics study on mir133 genes*. Thromb Res. 2015 Aug;136(2):367-70. IF=2.32
- Sandro Mancinelli, Francesca Lucaroni, Paola Borgiani, **Cinzia Ciccacci**, Leonardo Palombi, Patrizia De Filippis. *Low level exposure to arsenic in drinking water: a review on action mechanism, health effects and biomarkers*. IJSR 2015 Sept; 4 (9):191-97.
- Rufini Sara, **Ciccacci Cinzia**, Politi Cristina, Giardina Emiliano, Novelli Giuseppe, Borgiani Paola. *Stevens-Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN): an update on pharmacogenetics studies in drug-induced severe skin reaction*. Pharmacogenomics. 2015 Nov;16(17):1989-2002.
- Fulvia Ceccarelli, Carlo Perricone, Paola Borgiani, **Cinzia Ciccacci**, Sara Rufini, Enrica Cipriano, Cristiano Alessandri, Francesca Romana Spinelli, Antonio Sili Scavalli, Giuseppe Novelli, Guido Valesini, and Fabrizio Conti. *Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype*. Journal of Immunology Research 2015, 2015:745647.

- Ciccacci C**, Politi C, Novelli G, Borgiani P. *Advances In Exploring The Role Of Micrnas In Inflammatory Bowel Disease*. *Microna*. 2016, 5(1):5-11.
- Ciccacci C**, Conigliaro P, Perricone C, Rufini S, Triggianese P, Politi C, Novelli G, Perricone R, Borgiani P. *Polymorphisms in STAT4, IL10, PSORS1C1, PTPN2 and MIR146A genes are differently associated with prognostic factors in Italian patients affected by Rheumatoid Arthritis*. *Clin Exp Immunol*. 2016, 186(2):157-163.
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- Cristina Politi, **Cinzia Ciccacci**, Cinzia D'Amato, Giuseppe Novelli, Paola Borgiani, Vincenza Spallone. *Recent advances in exploring the genetic susceptibility to diabetic neuropathy*. *Diabetes Res Clin Pract*. 2016, 120:198-208.
- Ciccacci C**, Politi C, Biancone L, Latini A, Novelli G, Calabrese E, Borgiani P. *Polymorphisms in MIR122, MIR196A2, and MIR124A genes are associated with clinical phenotypes in Inflammatory Bowel Diseases*. *Mol Diagn Ther*. 2017, 21(1):107-114.
- Ciccacci C**, Perricone C, Politi C, Rufini S, Ceccarelli F, Cipriano E, Alessandri C, Latini A, Valesini G, Novelli G, Conti F, Borgiani P. *A polymorphism upstream MIR1279 gene is associated with pericarditis development in Systemic Lupus Erythematosus and contributes to definition of a genetic risk profile for this complication*. *Lupus* 2017, 26(8):841-848.
- Conigliaro P, **Ciccacci C**, Politi C, Triggianese P, Rufini S, Kroegler B, Perricone C, Latini A, Novelli G, Borgiani P, Perricone R. *Polymorphisms in STAT4, PTPN2, PSORS1C1 and TRAF3IP2 genes are associated with the response to TNF inhibitors in patients with Rheumatoid Arthritis*. *Plos One* 2017, 12(1): e0169956.
- Rufini S, **Ciccacci C**, Novelli G, Borgiani P. *Pharmacogenetics of Inflammatory Bowel Disease: a focus on Crohn's Disease*. *Pharmacogenomics* 2017, 18(11):1095-1114.
- Cinzia Ciccacci**, Andrea Latini, Cristina Politi, Sandro Mancinelli, Maria C. Marazzi, Giuseppe Novelli, Leonardo Palombi, Paola Borgiani. *Impact of Glutathione Transferases genes polymorphisms in Nevirapine adverse reactions: a possible role for GSTM1 in SJS/TEN susceptibility*. *Eur J Clin Pharm* 2017, 73 (10): 1253–1259.
- Andrea Latini, **Cinzia Ciccacci**, Giuseppe Novelli, Paola Borgiani. *Polymorphisms in miRNA genes and their involvement in autoimmune diseases susceptibility*. *Immunol Res*. 2017 Aug;65(4):811-827.
- Ciccacci C**, Latini A, Greco C, Politi C, D'Amato C, Lauro D, Novelli G, Borgiani P, Spallone V. *Association between a MIR499A polymorphism and diabetic neuropathy in type 2 diabetes*. *J Diabetes Complications*. 2018, 32 (1): 11-17.
- Politi C, **Ciccacci C**, Novelli G, Borgiani P. *Genetics and Treatment Response in Parkinson's Disease: An Update on Pharmacogenetic Studies*. *Neuromolecular Med*. 2018; 20(1):1-17
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**Ciccacci Cinzia**, Politi Cristina, Mancinelli Sandro, Ciccacci Fausto, Lucaroni Francesca, Novelli Giuseppe, Marazzi Maria Cristina, Palombi Leonardo, Borgiani Paola. *A multivariate genetic analysis confirms the rs5010528 in the HLA-C locus as a significant contributor to SJS/TEN susceptibility in Mozambique HIV population treated with Nevirapine*. *Journal of Antimicrobial Chemotherapy* 2018; 73 (8): 2137–2140

**Cinzia Ciccacci**, Carlo Perricone, Cristiano Alessandri, Andrea Latini, Cristina Politi, Federica Delunardo, Marina Pierdominici, Fabrizio Conti, Giuseppe Novelli, Elena Ortona, Paola Borgiani. *Evaluation of ATG5 polymorphisms in Italian patients with Systemic Lupus Erythematosus: contribution to disease susceptibility and clinical phenotypes*. *Lupus* 2018; 27(9) :1464-1469

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**C. Ciccacci**, A. Latini, C. Perricone, P. Conigliaro, S. Colafrancesco, F. Ceccarelli, R. Priori, F. Conti, R. Perricone, G. Novelli, and P. Borgiani. TNFAIP3 Gene Polymorphisms in Three Common Autoimmune Diseases: Systemic Lupus Erythematosus, Rheumatoid Arthritis, and Primary Sjogren Syndrome—Association with Disease Susceptibility and Clinical Phenotypes in Italian Patients. J Imm Res 2019, ID 6728694, doi.org/10.1155/2019/6728694

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**Ciccacci C**, Latini A, Colantuono A, Politi C, D'Amato C, Greco C, Rinaldi ME, Lauro D, Novelli G, Spallone V, Borgiani P. Expression study of candidate miRNAs and evaluation of their potential use as biomarkers of diabetic neuropathy. Epigenomics. 2020 Apr;12(7):575-585

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Autorizzo il trattamento dei miei dati personali ai sensi del GDPR e del Decreto Legislativo 30 giugno 2003, n. 196 “Codice in materia di protezione dei dati personali”