Cv Prof. Paola Borgiani

POSITON:
ASSOCIATE PROFESSOR OF MEDICAL GENETICS,
UNIVERSITY OF ROME “TOR VERGATA”

EDUCATION /TRAINING:
1980-1984: Degree in Biology, University of Camerino (Dissertation of a thesis in Human Genetics);
1987: PhD in Clinical and Biochemical Chemistry and Microbiology, University of Camerino (Dissertation of a thesis in Biochemical Genetics).
1983-85: Training in the Lab of Genetics, University of Camerino

PREVIOUS POSITIONS AND EMPLOYMENTS:

1985-1988: Research Assistant University of Camerino (Lab. Of Genetics), Dept Cellular Biology: Field of Study: Human Genetics

1989-2001: Assistant Professor University of Rome "Tor Vergata", School of Medicine (Pediatrics); Field of Study: Human Genetics

1st November 2001-31st October 2014: : Assistant Professor Researcher (Medical Genetics) University of Rome "Tor Vergata", School of Medicine.

1991: Visiting Researcher at the Lab. of Biochemical Genetics, Dept. Human Genetics, Children Hospital of Buffalo, Buffalo University (USA) for a collaborative project on genetic susceptibility to Diabetes and Obesity.

June 2000: Visiting Researcher at the Experimental Medicine Unit (University of Wales, UK) for a collaborative project on genetic susceptibility of allergy and athsma).

November 2004- April 2005: Secondment at the EMEA (European Medicine Agency) of London as “NATIONAL EXPERT” ” in Human Genetics. EMEA Safety and Efficacy of Medicine Sector,
(Field of Pharmacogenetics, Collaborates with Pharmacogenetics Working Party)

RESEARCH AREA:
Study of genetic factors and their interactions in the susceptibility to multifactorial diseases and their role in the clinical manifestations and phenotypes; Pharmacogenetics and Pharmacogenomics; Genomic Biomarkers and their application in the clinical practice. Personalized Medicine.
Co-author of more than 100 Papers published on “peer Reviewed” international Journal

2000-2015: Unit PI of about 10 different Grants Projects at national and international level

DIAGNOSTIC ACTIVITY
In the field of the molecular biology of the Achondroplasia, Galactosemia, and in the field of susceptibility genomic biomarkers, also in the Pharmacogenetics clinical applications.

TEACHING ACTIVITY:
Teaching Genetics, Medical Genetics, Pharmacogenomics at different courses of the Faculty of Medicine and Sciences, University of Rome, “Tor Vergata” (teaching Medical genetics at different PhD, Masters, degrees courses), also in English courses (i.e. Pharmacogenomics Couse at the European degree of Pharmacy, English degree in Medicine).

MEMBER of Italian Society of Human Genetics (SIGU): member of the Pharmacogenetics Working Party.

EDITORIAL BOARD MEMBER “Biomedicine and Prevention” http://www.biomedicineandprevention.com/)

Ad Hoc Reviewer for: Pharmacogenomics, PlosOne, Pharmacogenetics and Genomics,
Immunogenetics, Annals of Human Biology, Disease Markers, and other International Peer review Journals

Paola Borgiani: Pubblicazioni dal 2013 a Maggio 2017

- Polymorphisms in STAT4, PTPN2, PSORS1C1 and TRAF3IP2 Genes Are Associated with the Response to TNF Inhibitors in Patients with Rheumatoid Arthritis. Conigliaro P, Ciccacci C, Politi C, Triggianese P, Rufini S, Kroegler B, Perricone C, Latini A, Novelli G, **Borgiani P**, Perricone R. PLoS One. 2017 Jan 20;12(1):e0169956. doi: 10.1371/journal.pone.0169956. eCollection 2017.
- Genome-wide association study of nevirapine hypersensitivity in a sub-Saharan African HIV-infected population. Carr DF, Bourgeois S, Chaponda M, Takeshita LY, Morris AP, Castro EM, Alfirevic A, Jones AR, Rigden DJ, Haldenby S, Khoo S, Lalloo DG, Heyderman RS, Dandara C, Kampira E, van Oosterhout JJ, Ssali F, Munderi P, Novelli G, **Borgiani P**, Nelson MR, Holden A, Deloukas P, Pirmohamed M. J Antimicrob Chemother. 2017 Apr 1;72(4):1152-1162. doi: 10.1093/jac/dkw545.
- 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. Ciccacci C, Perricone C, Politi C, Rufini S, Ceccarelli F, Cipriano E, Alessandri C, Latini A, Valesini G, Novelli G, Conti F, **Borgiani P**. Lupus. 2016 Nov 22. pii: 0961203316679528. [Epub ahead of print]
- Polymorphisms in MIR122, MIR196A2, and MIR124A Genes are Associated with Clinical Phenotypes in Inflammatory Bowel Diseases. Ciccacci C, Politi C, Biancone L, Latini A, Novelli G, Calabrese E, **Borgiani P**.Mol Diagn Ther. 2017 Feb;21(1):107-114. doi: 10.1007/s40291-016-0240-1.
- A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. Mizzi C, Dalabira E, Kumuthini J, Dzimiri N, Balogh I, Başak N, Böhm R, Borg J, **Borgiani P**, Bozina N, Bruckmueller H, Burzynska B, Carracedo A, Cascorbi I, Deltas C, Dolzan V, Fenech A, Grech G, Kasiulevicius V, Kádaši Ľ, Kučinskas V, Khusnutdinova E, Loukas YL, Macek M Jr, Makukh H, Mathijssen R, Mitropoulos K, Mitropoulou C, Novelli G, Papantoni I, Pavlovic S, Saglio G, Setric J, Stojiljkovic M, Stubbs AP, Squassina A, Torres M, Turnovec M, van Schaik RH, Voskarides K, Wakil SM, Werk A, Del Zompo M, Zukic B, Katsila T, Lee MT, Motsinger-Rief A, Mc Leod HL, van der Spek PJ, Patrinos GP. PLoS One. 2016 Sep 16;11(9):e0162866. doi: 10.1371/journal.pone.0162866. eCollection 2016. Erratum in: PLoS One. 2017 Feb 16;12 (2):e0172595.
- Recent advances in exploring the genetic susceptibility to diabetic neuropathy. Politi C, Ciccacci C, D'Amato C, Novelli G, **Borgiani P**, Spallone V. Diabetes Res Clin Pract. 2016 Oct;120:198-208. doi: 10.1016/j.diabres.2016.08.006. Epub 2016 Aug 26. Review.
- Polymorphisms in STAT-4, IL-10, PSORS1C1, PTPN2 and MIR146A genes are associated differently with prognostic factors in Italian patients affected by rheumatoid arthritis. Ciccacci C, Conigliaro P, Perricone C, Rufini S, Triggianese P, Politi C, Novelli G, Perricone R, **Borgiani P**. Clin Exp Immunol. 2016 Nov;186(2):157-163. doi: 10.1111/cei.12831. Epub 2016 Aug 2.
- Cannabinoid Poisoning by Hemp Seed Oil in a Child. Chinello M, Scommegna S, Shardlow A, Mazzoli F, De Giovanni N, Fucci N, **Borgiani P**, Ciccacci C, Locasciulli A, Calvani M. Pediatr Emerg Care. 2016 Jun 14. [Epub ahead of print]
- 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 Jun;17(8):943-51. doi: 10.2217/pgs.16.20. Epub 2016 Jun 7.
- Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. Ceccarelli F, Perricone C, **Borgiani P**, Ciccacci C, Rufini S, Cipriano E, Alessandri C, Spinelli FR, Sili Scavalli A, Novelli G, Valesini G, Conti F. J Immunol Res. 2015;2015:745647. doi: 10.1155/2015/745647. Epub 2015 Dec 21. Review.
- Advances in Exploring the Role of Micrornas in Inflammatory Bowel Disease. Ciccacci C, Politi C, Novelli G, **Borgiani P**.Microrna. 2016;5(1):5-11.
- 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. 2016 Mar;37(5-6):860-4. doi: 10.1002/elps.201500346. Epub 2016 Jan 15.
- Stevens-Johnson syndrome and toxic epidermal necrolysis: an update on pharmacogenetics studies in drug-induced severe skin reaction. Rufini S, Ciccacci C, Politi C, Giardina E, Novelli G, **Borgiani P**. Pharmacogenomics. 2015 Nov;16(17):1989-2002. doi: 10.2217/pgs.15.128. Epub 2015 Nov 10. Review.
- 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 Sep;73(3):528-9. doi: 10.1016/j.jaad.2015.06.022. No abstract available.
- 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. Epub 2015 Jul 31.
- Could MicroRNA polymorphisms influence warfarin dosing? A pharmacogenetics study on mir133 genes. Ciccacci C, Rufini S, Politi C, Novelli G,Forte V, **Borgiani P** Thromb Res. 2015 Aug;136(2):367-70. doi: 10.1016/j.thromres.2015.06.026. Epub 2015 Jun 18.

- Autophagy and inflammatory bowel disease: Association between variants of the autophagy-related IRGM gene and susceptibility to Crohn's disease. Rufini S, Ciccacci C, Di Fusco D, Ruffa A, Pallone F, Novelli G, Biancone L, **Borgiani P**. Dig Liver Dis. 2015 Sep;47(9):744-50. doi: 10.1016/j.dld.2015.05.012. Epub 2015 May 21.
- A pharmacogenetics study in Mozambican patients treated with nevirapine: full resequencing of TRAF3IP2 gene shows a novel association with SJS/TEN susceptibility.Ciccacci C, Rufini S, Mancinelli S, Buonomo E, Giardina E, Scarcella P, Marazzi MC, Novelli G, Palombi L, **Borgiani P**. Int J Mol Sci. 2015 Mar 12;16(3):5830-8. doi: 10.3390/ijms16035830.
- Crohn's disease, the mycobacterium paratuberculosis and the genetic bond: An unexpected trio. Perricone C, **Borgiani P**. Clin Res Hepatol Gastroenterol. 2015 Jun;39(3):275-7. doi: 10.1016/j.clinre.2015.02.001. Epub 2015 Mar 11. No abstract available.
- 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. Epub 2014 Nov 12. Review.
- A multilocus genetic study in a cohort of Italian SLE patients confirms the association with STAT4 gene and describes a new association with HCP5 gene. Ciccacci C, Perricone C, Ceccarelli F, Rufini S, Di Fusco D, Alessandri C, Spinelli FR, Cipriano E, Novelli G, Valesini G, **Borgiani P**, Conti F. PLoS One. 2014 Nov 4;9(11):e111991. doi: 10.1371/journal.pone.0111991. eCollection 2014.
- 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. 2015 Apr;15(2):196-200. doi: 10.1038/tpj.2014.48. Epub 2014 Sep 9.
- Common polymorphisms in MIR146a, MIR128a and MIR27a genes contribute to neuropathy susceptibility in type 2 diabetes. 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. Acta Diabetol. 2014 Mar 30. [Epub ahead of print]
- A family study of asymptomatic small bowel Crohn's disease. Biancone L, Calabrese E, Petruzziello C, Capanna A, Zorzi F, Onali S, Condino G, Lolli E, Ciccacci C,**Borgiani P**, Pallone F. Dig Liver Dis. 2013 Dec 17. pii: S1590-8658(13)00656-7. doi: 10.1016/j.dld.2013.11.003. [Epub ahead of print]
- HCP5 genetic variant (RS3099844) contributes to Nevirapine-induced Stevens Johnsons Syndrome/Toxic Epidermal Necrolysis susceptibility in a population from Mozambique. **Borgiani P**, Di Fusco D, Erba F, Marazzi MC, Mancinelli S, Novelli G, Palombi L, Ciccacci C. Eur J Clin Pharmacol. 2013 Dec 10. [Epub ahead of print]
- High warfarin sensitivity in carriers of CYP2C9*35 is determined by the impaired interaction with P450 oxidoreductase. Lee MY, **Borgiani P**, Johansson I, Oteri F, Mkrtchian S, Falconi M, Ingelman-Sundberg M. Pharmacogenomics J. 2013 Dec 10. doi: 10.1038/tpj.2013.41. [Epub ahead of print]
- 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 Jun 19;8(6):e66978. Print 2013.
- TRAF3IP2 gene and systemic lupus erythematosus: association with disease susceptibility and pericarditis development. Perricone C, Ciccacci C, Ceccarelli F, Di Fusco D, Spinelli FR, Cipriano E, Novelli G, Valesini G, Conti F, **Borgiani P**. Immunogenetics. 2013 Oct;65(10):703-9. doi: 10.1007/s00251-013-0717-6. Epub 2013 Jul 9.
- Association between CYP2B6 polymorphisms and Nevirapine-induced SJS/TEN: a pharmacogenetics study. Ciccacci C, Di Fusco D, Marazzi MC, Zimba I, Erba F, Novelli G, Palombi L, **Borgiani P**, Liotta G.Eur J Clin Pharmacol. 2013 Nov;69(11):1909-16. doi: 10.1007/s00228-013-1549-x. Epub 2013 Jun 18.
- Resequencing of VKORC1, CYP2C9 and CYP4F2 genes in Italian patients requiring extreme low and high warfarin doses. Di Fusco D, Ciccacci C, Rufini S, Forte V, Novelli G, **Borgiani P**. Thromb Res. 2013 Jul;132(1):123-6. doi: 10.1016/j.thromres.2013.05.002. Epub 2013 May 30.
- MicroRNA genetic variations: association with type 2 diabetes. Ciccacci C, Di Fusco D, Cacciotti L, Morganti R, D'Amato C, Greco C, Rufini S, Novelli G, Sangiuolo F, Spallone V, **Borgiani P**. Acta Diabetol. 2013 Dec;50(6):867-72. doi: 10.1007/s00592-013-0469-7. Epub 2013 Mar 27.
- ABCC10 rs2125739 polymorphism and nevirapine-induced hepatotoxicity: lack of association in a population from Mozambique. Ciccacci C, Di Fusco D, Marazzi MC, Liotta G, Palombi L, Novelli G, **Borgiani P**. Pharmacogenet Genomics. 2013 Jan;23(1):38-9. doi: 10.1097/FPC.0b013e328359e951.