

DIDACTIC-SCIENTIFIC CURRICULUM OF PROF. EMILIANO GIARDINA

PERSONAL DATA

Name and Surname: Emiliano Giardina

Place and date of birth: Rome, 16-04-1976

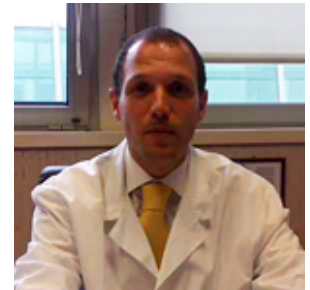
Current position: Professor of Medical and Forensic Genetics at the University of Rome "Tor Vergata"

Department: Biomedicine and Prevention

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SCIENTIFIC AND DIDACTIC ACTIVITY

Academic and educational qualifications:

2015: Associate Professor in Medical Genetics (sector MED/03)

2010: Specialization Degree in Medical Genetics

2006: Phd in Cellular Death Physiopathology

2004: University Researcher in the Disciplinary Scientific Sector MED/03

2000: Degree certificate in Biological Sciences

BIBLIOMETRIC PARAMETERS

Number of scientific publications >80

H index: 26

Number of citations: 3126

Professional and teaching activities:

- Director of the Forensic Genetics Laboratory in the University of Rome "Tor Vergata".
- Director of the Genomic Medicine Laboratory of UILDM in Santa Lucia Foundation in Rome.
- Director of the II Level Master Degree in "Forensic Genetics" in the University of Rome "Tor Vergata",
- Director and Founder of the Permanent School of Forensic Biology instituted at the University of Rome "Tor Vergata" and funded by the National Association of Biologists
- National Coordinator of the Forensic Genetics Work Group of Italian Society of Human Genetics
- Permanent Member of the group interdisciplinary for the Italian National DNA Database.
- Head of Forensic Biology section at the Italian Association of Biologists
- Lecturer of Medical Genetics of the Master Degree in Medicine and Surgery at the University of Rome "Tor Vergata".
- Lecturer of Medical Genetics of the Master Degrees in "Medical Biotechnology" at the University of Rome "Tor Vergata".
- Lecturer of "Biology and Human Evolution" at the University of Rome "Tor Vergata".
- Lecturer of Medical Genetics of the Bachelor Degree in "Biotechnology" at the University of Urbino "Carlo Bo".
- Lecturer of Optogenetics of the Ophthalmology Specialization School at the University of Rome "Tor Vergata".

- Lecturer of Medical Genetics of the Nephrology Specialization School at the University of Rome “Tor Vergata”.
- Lecturer of “Immunology and Applied Biotechnologies” PhD and “Advanced Technologies in Biomedicine” PhD in the University of Rome “Tor Vergata”.

Scientific activity (2001-2017):

- Member of the research group of the “Excellence Centre for the study of genomic risk in multifactorial complex Diseases” in the University of Rome “Tor Vergata”.
- Participation to the International Consortium for the study of Genetic Susceptibility to Psoriasis.
- Participation to the COFIN 2003 Project “Study of the Expression of the genes mapped in the PSORS1 and PSORS4 regions and other genes involved in psoriasis pathogenesis”.
- Participation to the “NACBO” European Project concerning the application of nano-biotechnologies in health-care and forensic fields.
- Collaboration with the Forensic Medicine department of the University of Rome “Tor Vergata” to develop projects in the field of forensic genetics.
- Collaboration with CNR for the “Study of the Molecular Recognition Chemistry”, concerning the computational prediction of the impact of genetic variants on the protein folding.

Awards for research activity: “Youth Knighthood” Award for the scientific contributions offered in the field of medical genetics (2009).

RESEARCH ACTIVITY:

1. Capon F, Tacconelli A, Giardina E, Sciacchitano S, Bruno R, Trischitta V, Tassi V, Filetti S, Dallapiccola B, Novelli G. Mapping a dominant form of multinodular goiter to chromosome Xp22. *Am J Hum Genet.* 2000 Oct;67(4):1004-7.
2. Giardina E, Capon F, D'Apice MR, Amati F, Arturi F, Filetti S, Bonifazi E, Pucci S, Conte C, Novelli G. Mutational analysis of Peroxiredoxin IV: exclusion of a positional candidate for multinodular goitre. *BMC Medical Genetics* 2002, 23 July, 3:5.
3. Semprini S, Capon F, Tacconelli A, Giardina E, Orecchia A, Mingarelli R, Gobello T, Zambruno G, Botta A, Fabrizi G, Novelli G. Evidence for differential S100 gene over-expression in psoriatic patients from genetically heterogeneous pedigrees. *Hum Genet.* 2002 Oct;111(4-5):310-3
4. Borgiani P, Vallo L, D'Apice MR, Giardina E, Pucci S, Capon F, Nistic S, Chimenti S, Pallone F, Novelli G. Exclusion of CARD15/NOD2 as a candidate susceptibility gene to psoriasis in the Italian population. *Eur J Dermatol.* 2002 Nov-Dec;12(6): 540-2
5. Novelli G, Borgiani P, Giardina E, Mango R, Contino G, Romeo F, Mehta JL. Role of genetics in prevention of coronary atherosclerosis. *Curr Opin Cardiol.* 2003 Sep;18(5):368-371.
6. Mango R, Clementi F, Borgiani P, Forleo GB, Federici M, Contino G, Giardina E, Garza L, Fahdi IE, Lauro R, Mehta JL, Novelli G, Romeo F. Association of single nucleotide polymorphisms in the oxidised LDL receptor 1(OLR1) gene in patients with acute myocardial infarction. *J Med Genet.* 2003 Dec;40(12):933-6.
7. Novelli, G., Giardina, E., Paradisi, M., Pedicelli, C., Girolomoni, G., Nasorri, F., et al. (2003). Insight into genetics of atopic dermatitis: Future approaches and directions. *Journal of Investigative Dermatology*, 121(5), 1265-1265.
8. E. Giardina, F. Capon, MC. DE Rosa, R. Mango, G. Zambruno, A. Orecchia, S. Chimenti, B. Giardina, G. Novelli. Characterization of the Loricrin (LOR) Gene as a Positional Candidate for the PSORS4 Psoriasis Susceptibility locus. *Ann Hum Genet.* 2004 Nov;68(Pt 6):639-45
9. E. Giardina, G. Novelli, A. Costanzo, S. Nistico, C. Bulli, C. Sinibaldi, M.L. Sorgi, S. Chimenti, F. Pallone, E. Taccari, P. Borgiani. Psoriatic Arthritis and CARD15 Gene Polymorphisms: No Evidence for Association in the Italian Population. *J Invest Dermatol* 2004 May;122(5):1106-7

10. E. Giardina, C. Sinibaldi, G. Novelli. The Psoriasis Genetics as a Model of Complex Disease. *Curr Drug Targets Inflamm Allergy*, 2004, 3, 129-136. Jun;3(2):129-36.
11. Emanuela Bonifazi, Laura Vallo, Emiliano Giardina, Annalisa Botta and Giuseppe Novelli. A Long PCR-Based Molecular Protocol for Detecting Normal and Expanded ZNF9 Alleles in Myotonic Dystrophy Type 2. *Diagn Mol Pathos*, 2004 Sep;13(3):164-166.
12. Giuseppe Novelli, Emiliano Giardina. The Genetics of Psoriasis. In *Recent Research Developments in Genetics; Research Signpost*, T. C. 37/661(2), Fort Post Office, Trivandrum - 695023, Kerala, India.
13. Sangiuolo F, Filareto A, Giardina E, Nardone AM, Pilu G, Pietropolli A, Bertini E, Novelli G. Prenatal diagnosis of spinal muscular atrophy with respiratory distress (SMARD1) in a twin pregnancy. *Prenat Diagn*, 2004 Oct;24(10):839-41.
14. Elder JT; Cluster 17 Collaboration. Fine mapping of the psoriasis susceptibility gene PSORS1: a reassessment of risk associated with a putative risk haplotype lacking HLA-Cw6. *J Invest Dermatol*, 2005 May; 124(5):921-30.
15. Capon F and Giardina E. The Long and winding road: searching for non-MHC psoriasis Susceptibility Loci. *Curr Genomics*, 2005; (6): 45-49.
16. Botta A, Tacconelli A, Bagni I, Giardina E, Bonifazi E, Pietropolli A, Clementi M, Novelli G. Transmission ratio distortion in the spinal muscular atrophy locus: data from 314 prenatal tests. *Neurology*, 2005; 65(10):1631-1635.
17. Concolino P, Satta MA, Santonocito C, Carrozza C, Rocchetti S, Ameglio F, Giardina E, Zuppi C, Capoluongo E. Linkage between I172N mutation, a marker of 21-hydroxylase deficiency, and a single nucleotide polymorphism in Int6 of CYP21B gene: A genetic study of Sardinian family. *Clin Chim Acta*, 2006; 364(1-2):298-302.
18. Giardina E, Predazzi I, Sinibaldi C, Peconi C, Amerio P, Costanzo A, Paradisi A, Capizzi R, Paradisi M, Chimenti S, Taccari E, Novelli G. PSORS2 markers are not associated with psoriatic arthritis in the Italian population. *Hum Hered*, 2006; 61(2):120-122.
19. Giardina E, Sinibaldi C, Chini L, Moschese V, Marulli G, Provini A, Rossi P, Paradisi M, Chimenti S, Galli E, Brunetti E, Girolomoni G, Novelli G. Co-localization of susceptibility loci for psoriasis (PSORS4) and atopic dermatitis (ATOD2) on human chromosome 1q21. *Hum Hered*, 2006; 61(4):229-236.
20. Guarino S, Perricone C, Guarino MD, Giardina E, Gambardella S, Rosaria D'Apice M, Bulli C, Perricone R, Novelli G. Gonadal mosaicism in hereditary angioedema. *Clin Genet*, 2006; 70(1):83-85.
21. Porzio O, Cunsolo V, Malaponti M, De Nisco E, Acquafredda A, Cavallo L, Andreani M, Giardina E, Testi M, Cappa M, Federici G. Divergent phenotype of two siblings HLA identical, affected by nonclassical and classical CAH caused by 21-Hydroxylase deficiency. *J Clin Endocrinol Metab*, 2006; 91(11):4510-4513.
22. Giardina E, Sinibaldi C and Giuseppe Novelli. Mapping the future of common diseases: lesson from psoriasis. *Front Biosci*, 2007; 12:1563-1573.
23. Capoluongo E, Vento G, Rocchetti S, Giardina E, Concolino P, Sinibaldi C, Santonocito C, Vendettuoli V, Tana M, Tirone C, Zuppi C, Romagnoli C, Novelli G, Giardina B, Ameglio F. Mannose-binding lectin polymorphisms and pulmonary outcome in premature neonates: a pilot study. *Intensive Care Med*. 2007; 33(10):1787-94.
24. Giardina E, Pietrangeli I, Martone C, Asili P, Predazzi I, Marsala P, Gabriele L, Pipolo C, Ricci O, Solla G, Sineo L, Spinella A, Novelli G. In silico and in vitro comparative analysis to select, validate and test SNPs for human identification. *BMC Genomics*. 2007 Dec 12;8(1):457
25. Giardina E, Paolillo N, Sinibaldi C, Novelli G. R501X and 2282del4 filaggrin mutations do not confer susceptibility to psoriasis and atopic dermatitis in Italian patients. *Dermatology*. 2008;216(1):83-4.
26. Giardina E, Predazzi I, Pietrangeli I, Asili P, Marsala P, Gabriele L, Pipolo C, Ricci O, Martone C, Spinella A, Novelli G. Frequency assessment of SNPs for forensic identification in different populations. *FSIGEN* 2007 Dec;1(3-4):e1-3.

27. Giardina E, Pietrangeli I, Martínez-Labarga C, Martone C, De Angelis F, Spinella A, De Stefano G, Rickards O, Novelli G. Haplotypes in SLC24A5 gene as Ancestry Informative Markers in different populations. *Curr Genomics* 2008.
28. Giardina E, Peconi C, Cascella R, Sinibaldi C, Nardone A,M, Novelli G.. A multiplex molecular assay for the detection of uniparental disomy (UPD) for human chromosome 15. *Electrophoresis*. 2008 Dec;29(23):4775-9.
29. de Cid R, Riveira-Munoz E, Zeeuwen PL, Robarge J, Liao W, Dannhauser EN, Giardina E, Stuart PE, Nair R, Helms C, Escaramís G, Ballana E, Martín-Ezquerria G, Heijer MD, Kamsteeg M, Joosten I, Eichler EE, Lázaro C, Pujol RM, Armengol L, Abecasis G, Elder JT, Novelli G, Armour JA, Kwok PY, Bowcock A, Schalkwijk J, Estivill X. Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. *Nat Genet*. 2009 Feb;41(2):211-215.
30. Bellia A, Giardina E, Lauro D, Tesauro M, Di Fede G, Cusumano G, Federici M, Rini GB, Novelli G, Lauro R, Sbraccia P. "The Linosa Study": Epidemiological and heritability data of the metabolic syndrome in a Caucasian genetic isolate. *Nutr Metab Cardiovasc Dis*. 2009 Feb 5.
31. Giardina E, Pietrangeli I, Martone C, Zampatti S, Marsala P, Gabriele L, Ricci O, Solla G, Asili P, Arcudi G, Spinella A, Novelli G. Whole genome amplification and real-time PCR in forensic casework. *BMC Genomics*. 2009 Apr 14;10(1):159.
32. Pietrangeli I, Caruso V, Veneziano L, Spinella A, Arcudi G, Giardina E, Novelli G. Forensic DNA challenges: replacing numbers with names of Fosse Ardeatine's victims. *J Forensic Sci*. 2009 Jul;54(4):905-8.
33. Giardina E, Peconi C, Cascella R, Sinibaldi C, Foti Cuzzola V, Nardone AM, Bramanti P, Novelli G. A multiplex molecular assay for the detection of uniparental disomy for human chromosome 7. *Electrophoresis*. 2009 Jun;30 (11):2008-11.
34. Concolino P, Mello E, Minucci A, Giardina E, Zuppi C, Toscano V, Capoluongo E. A new CYP21A1P/CYP21A2 chimeric gene identified in an Italian woman suffering from classical congenital adrenal hyperplasia form. *BMC Med Genet*. 2009 Jul 22;10:72.
35. Ricci F, Zampatti S, D'Abbruzzi F, Missiroli F, Martone C, Lepre T, Pietrangeli I, Sinibaldi C, Peconi C, Novelli G, Giardina E. Typing of ARMS2 and CFH in age-related macular degeneration: case-control study and assessment of frequency in the Italian population. *Arch Ophthalmol*. 2009 Oct;127(10):1368-72.
36. Chiriaco M, Di Matteo G, Sinibaldi C, Giardina E, Nardone AM, Folgore L, D'Argenio P, Rossi P, Finocchi A. Identification of Deletion Carriers in X-Linked Chronic Granulomatous Disease by Real-Time PCR. *Genet Test Mol Biomarkers*. 2009 Oct 19.
37. Spitalieri P, Cortese G, Pietropolli A, Filareto A, Dolci S, Klinger FG, Giardina E, Di Cesare S, Bernardini L, Lauro D, Scaldaferrì HL, Citro G, Novelli G, De Felici M, Sangiuolo F. Identification of multipotent cytotrophoblast cells from human first trimester chorionic villi. *Cloning Stem Cells*. 2009 Dec;11(4):535-56.
38. Bergboer JG, Zeeuwen PL, Irvine AD, Weidinger S, Giardina E, Novelli G, Heijer MD, Rodriguez E, Illig T, Riveira-Munoz E, Campbell LE, Tyson J, Dannhauser EN, O'Regan GM, Galli E, Klopp N, Koppelman GH, Novak N, Estivill X, McLean WH, Postma DS, Armour JA, Schalkwijk J. Deletion of Late Cornified Envelope 3B and 3C Genes Is Not Associated with Atopic Dermatitis. *J Invest Dermatol*. 2010 Apr 8.
39. Pietrangeli I, Ottaviani E, Martone C, Gabriele L, Arcudi G, Potenza S, Spinella A, Giardina E, Novelli G. Frequency assessment of 25 SNPs in five different populations. *Forensic Sci Int Genet*. 2010 Oct;4(5):e131-3. IF= 2.421
Pietrangeli I, Ottaviani E, Martone C, Gabriele L, Arcudi G, Potenza S, Spinella A, Giardina E, Novelli G. Frequency assessment of 25 SNPs in five different populations. *Forensic Sci Int Genet*. 2010 Oct;4(5):e131-3. IF= 2.421
40. Giardina E, Stocchi L, Cuzzola VF, Zampatti S, Gambardella S, Patrizi MP, Bramanti P, Pirazzoli A, Novelli G. A fluorescence-based sequence-specific primer PCR for the screening of HLA-B(*)57:01. *Electrophoresis*. 2010 Oct 5. IF= 3.609

41. Ulrike Hüffmeier, Steffen Uebe, Arif B. Ekici, John Bowes, Emiliano Giardina, Eleanor Korendowych, Kristina Juneblad, Maria Apel, Ross McManus, Pauline Ho, Ian N. Bruce, Anthony W. Ryan, Frank Behrens, Jesús Lascorz, Beate Böhm, Heiko Traupe, Jörg Lohmann, Christian Gieger, Heinz-Erich Wichmann, Christine Herold, Michael Steffens, Lars Klareskog, Thomas F. Wienker, Oliver FitzGerald, Gerd-Marie Alenius, Neil J. McHugh, Giuseppe Novelli, Harald Burkhardt, Anne Barton, André Reis. Missense variant in TRAF3IP2 associates with psoriatic arthritis and psoriasis. *Nat Genet.* in press IF= 34.284
42. Amy Strange, Francesca Capon, Chris CA Spencer, Jo Knight, Michael E Weale, Michael H Allen, Anne Barton, Gavin Band, Céline Bellenguez, Judith GM Bergboer, Jenefer M Blackwell, Elvira Bramon, Suzannah J Bumpstead, Juan P Casas, Michael J Cork, Aiden Corvin, Panos Deloukas, Serge Dronov, Audrey Duncanson, Sarah Edkins, Xavier Estivill, Oliver Fitzgerald, Colin Freeman, Emiliano Giardina, Emma Gray, Angelika Hofer, Ulrike Hüffmeier, Sarah E Hunt, Alan D Irvine, Janusz Jankowski, Brian Kirby, Cordelia Langford, Jesús Lascorz, Joyce Leman, Lotus Mallbris, Hugh S Markus, Christopher G Mathew, WH Irwin McLean, Ross McManus, Rotraut Mössner, Åsa T Naluai, Frank O Nestle, Giuseppe Novelli, Alexandros Onoufriadis, Colin NA Palmer, Carlo Perricone, Matti Pirinen, Robert Plomin, Ramon M Pujol, Anna Rautanen, Eva Riveira-Munoz, Anthony W Ryan, Wolfgang Salmhofer, Lena Samuelsson, Stephen J Sawcer, Joost Schalkwijk, Catherine H Smith, Mona Stähle, Rachid Tazi-Ahnini, Heiko Traupe, Ananth C Viswanathan, Richard B Warren, Wolfgang Weger, Katarina Wolk, Nicholas Wood, Jane Worthington, Helen S Young, Patrick LJM Zeeuwen, Adrian Hayday, A David Burden, Christopher EM Griffiths, Juha Kere, André Reis, David Evans, Matthew A Brown, Jonathan N Barker, Leena Peltonen, Peter Donnelly and Richard C Trembath. Identification of novel psoriasis susceptibility loci and genetic interaction between HLA-C and ERAP1 provides evidence for an integrated pathogenic pathway *Nat Genet.* in press IF= 34.284
43. Riveira-Munoz E, He SM, Escaramís G, Stuart PE, Hüffmeier U, Lee C, Kirby B, Oka A, Giardina E, Liao W, Bergboer J, Kainu K, de Cid R, Munkhbat B, Zeeuwen PL, Armour JA, Poon A, Mabuchi T, Ozawa A, Zawirska A, Burden AD, Barker JN, Capon F, Traupe H, Sun LD, Cui Y, Yin XY, Chen G, Lim HW, Nair RP, Voorhees JJ, Tejasvi T, Pujol R, Munkhtuvshin N, Fischer J, Kere J, Schalkwijk J, Bowcock A, Kwok PY, Novelli G, Inoko H, Ryan AW, Trembath RC, Reis A, Zhang XJ, Elder JT, Estivill X. Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. *J Invest Dermatol.* 2010 Nov 25. IF= 5.543
44. Nisticò S, Paolillo N, Minella D, Piccirilli S, Rispoli V, Giardina E, Biancolella M, Chimenti S, Novelli G, Nisticò G. Effects of TNF-alpha and IL-1beta on the activation of genes related to inflammatory, immune responses and cell death in immortalized human HaCat keratinocytes. *Int J Immunopathol Pharmacol.* 2010 Oct-Dec;23(4):1057-72. IF= 2.793
45. Cascella R, Cuzzola VF, Lepre T, Galli E, Moschese V, Chini L, Mazzanti C, Fortugno P, Novelli G, Giardina E. Full Sequencing of the FLG Gene in Italian Patients with Atopic Eczema: Evidence of New Mutations, but Lack of an Association. *J Invest Dermatol.* 2011 Feb 3. IF= 5.543
46. Giardina E, Spinella A, Novelli G. Past, present and future of forensic DNA typing. *Nanomedicine (Lond).* 2011 Feb;6(2):257-70. Review. IF= 6.2
47. Docampo E, Giardina E, Riveira-Muñoz E, de Cid R, Escaramís G, Perricone C, Fernández-Sueiro JL, Maymó J, González-Gay MA, Blanco FJ, Hüffmeier U, Lisbona MP, Martín J, Carracedo A, Reis A, Rabionet R, Novelli G, Estivill X. Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: a study in Spanish and Italian populations and meta-analysis. *Arthritis Rheum.* 2011 Jul;63(7):1860-5. IF= 8.4
48. Lepre T, Cascella R, Missiroli F, De Felici C, Taglia F, Zampatti S, Cusumano A, Ricci F, Giardina E, Eandi CM, Novelli G. Polymorphisms in ARMS2 (LOC387715) and LOXL1 genes in the Japanese with age-related macular degeneration. *Am J Ophthalmol.* 2011 Aug;152(2):325-6. IF= 3.8
49. Giardina E, Hüffmeier U, Ravindran J, Behrens F, Lepre T, McHugh NJ, Korendowych E, Burkhardt H, Novelli G, Reis A. Tumor necrosis factor promoter polymorphism TNF*-857 is a

- risk allele for psoriatic arthritis independent of the PSORS1 locus. *Arthritis Rheum.* 2011 Dec;63(12):3801-6. IF= 8.4
50. Paolillo N, Piccirilli S, Giardina E, Rispoli V, Colica C, Nisticò S. Effects of paraquat and capsaicin on the expression of genes related to inflammatory, immune responses and cell death in immortalized human HaCat keratinocytes. *Int J Immunopathol Pharmacol.* 2011 Oct-Dec;24(4):861-8. IF= 3.6
 51. Pigionica M, Baldassarra SL, Giardina E, Stella A, D'Ovidio FD, Frati P, Lenato GM, Resta N, Dell'erba A. Population data for 17 Y-chromosome STRs in a sample from Apulia (Southern Italy). *Forensic Sci Int Genet.* 2012 Sep 6.
 52. Previderè C, Grignani P, Alessandrini F, Alù M, Biondo R, Boschi I, Caenazzo L, Carboni I, Carnevali E, De Stefano F, Domenici R, Fabbri M, Giardina E, Inturri S, Pelotti S, Piccinini A, Pigionica M, Resta N, Turrina S, Verzeletti A, Presciuttini S. The 2011 GeFI collaborative exercise. Concordance study, proficiency testing and Italian population data on the new ENFSI/EDNAP loci D1S1656, D2S441, D10S1248, D12S391, D22S1045. *Forensic Sci Int Genet.* 2012 Aug 20.
 53. 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.* 2012 Mar 23.
 54. Tsoi LC, Spain SL, Knight J, Ellinghaus E, Stuart PE, Capon F, Ding J, Li Y, Tejasvi T, Gudjonsson JE, Kang HM, Allen MH, McManus R, Novelli G, Samuelsson L, Schalkwijk J, Ståhle M, Burden AD, Smith CH, Cork MJ, Estivill X, Bowcock AM, Krueger GG, Weger W, Worthington J, Tazi-Ahnini R, Nestle FO, Hayday A, Hoffmann P, Winkelmann J, Wijmenga C, Langford C, Edkins S, Andrews R, Blackburn H, Strange A, Band G, Pearson RD, Vukcevic D, Spencer CC, Deloukas P, Mrowietz U, Schreiber S, Weidinger S, Koks S, Kingo K, Esko T, Metspalu A, Lim HW, Voorhees JJ, Weichenthal M, Wichmann HE, Chandran V, Rosen CF, Rahman P, Gladman DD, Griffiths CE, Reis A, Kere J; Collaborative Association Study of Psoriasis (CASP), Duffin KC, Helms C, Goldgar D, Li Y, Paschall J, Malloy MJ, Pullinger CR, Kane JP, Gardner J, Perlmutter A, Miner A, Feng BJ, Hiremagalore R, Ike RW, Christophers E, Henseler T, Ruether A, Schrodi SJ, Prahalad S, Guthery SL, Fischer J, Liao W, Kwok P, Menter A, Lathrop GM, Wise C, Begovich AB; Genetic Analysis of Psoriasis Consortium, Onoufriadis A, Weale ME, Hofer A, Salmhofer W, Wolf P, Kainu K, Saarialho-Kere U, Suomela S, Badorf P, Hüffmeier U, Kurrat W, Küster W, Lascorz J, Mössner R, Schürmeier-Horst F, Ständer M, Traupe H, Bergboer JG, Heijer MD, van de Kerkhof PC, Zeeuwen PL, Barnes L, Campbell LE, Cusack C, Coleman C, Conroy J, Ennis S, Fitzgerald O, Gallagher P, Irvine AD, Kirby B, Markham T, McLean WH, McPartlin J, Rogers SF, Ryan AW, Zawirska A, Giardina E, Lepre T, Perricone C, Martín-Ezquerria G, Pujol RM, Riveira-Munoz E, Inerot A, Nalwai AT, Mallbris L, Wolk K, Leman J, Barton A, Warren RB, Young HS, Ricano-Ponce I, Trynka G; Psoriasis Association Genetics Extension, Pellett FJ, Henschel A, Aurand M, Bebo B, Gieger C, Illig T, Moebus S, Jöckel KH, Erbel R; Wellcome Trust Case Control Consortium 2, Donnelly P, Peltonen L, Blackwell JM, Bramon E, Brown MA, Casas JP, Corvin A, Craddock N, Duncanson A, Jankowski J, Markus HS, Mathew CG, McCarthy MI, Palmer CN, Plomin R, Rautanen A, Sawcer SJ, Samani N, Viswanathan AC, Wood NW, Bellenguez C, Freeman C, Hellenthal G, Giannoulatou E, Pirinen M, Su Z, Hunt SE, Gwilliam R, Bumpstead SJ, Dronov S, Gillman M, Gray E, Hammond N, Jayakumar A, McCann OT, Liddle J, Perez ML, Potter SC, Ravindrarajah R, Ricketts M, Waller M, Weston P, Widaa S, Whittaker P, Nair RP, Franke A, Barker JN, Abecasis GR, Elder JT, Trembath RC. Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nat Genet.* 2012 Nov 11.
 55. Pigionica M, Loner Baldassarra S, Giardina E, Tonino Marsella L, Resta N, Dell'erba A. Allele frequencies of the new European Standard Set (ESS) loci in a population of Apulia (Southern Italy). *Forensic Sci Int Genet.* 2012 Nov 2.

56. Stocchi L, Cascella R, Zampatti S, Pirazzoli A, Novelli G, Giardina E. The Pharmacogenomic HLA Biomarker Associated to Adverse Abacavir Reactions: Comparative Analysis of Different Genotyping Methods. *Curr Genomics*. 2012 Jun;13(4):314-20.
57. Apel M, Uebe S, Bowes J, Giardina E, Korendowych E, Juneblad K, Pasutto F, Ekici AB, McManus R, Ho P, Bruce IN, Ryan AW, Behrens F, Böhm B, Traupe H, Lohmann J, Gieger C, Wichmann HE, Padyukov L, Fitzgerald O, Alenius GM, McHugh NJ, Novelli G, Burkhardt H, Barton A, Reis A, Hüffmeier U. Variants in RUNX3 contribute to susceptibility to psoriatic arthritis, exhibiting further common ground with ankylosing spondylitis. *Arthritis Rheum*. 2013 May;65(5):1224-31.
58. Ricci F, Staurengi G, Lepre T, Missiroli F, Zampatti S, Cascella R, Borgiani P, Marsella LT, Eandi CM, Cusumano A, Novelli G, Giardina E. Haplotypes in IL-8 Gene Are Associated to Age-Related Macular Degeneration: A Case-Control Study. *PLoS One*. 2013 Jun 19;8(6)
59. Pietropolli A, Vicario R, Peconi C, Zampatti S, Quitadamo MC, Capogna MV, Ragazzo M, Nardone AM, Postorivo D, Spitalieri P, Sarta S, Ratto F, Novelli G, Sangiuolo F, Piccione E, Giardina E. Transabdominal coelocentesis as early source of fetal DNA for chromosomal and molecular diagnosis. *J Matern Fetal Neonatal Med*. 2014 Apr 9.
60. Porter C, Giardina E, Eusebi L. Clinical trial sponsors' refusal to communicate genetic research results to subjects. *Patient Educ Couns*. 2014 Apr;95(1):157-8.
61. Zampatti S, Ricci F, Cusumano A, Marsella LT, Novelli G, Giardina E. Review of nutrient actions on age-related macular degeneration. *Nutr Res*. 2014 Feb;34(2):95-105.
62. Giardina E, Oddone F, Lepre T, Centofanti M, Peconi C, Tanga L, Quaranta L, Frezzotti P, Novelli G, Manni G. Common sequence variants in the LOXL1 gene in pigment dispersion syndrome and pigmentary glaucoma. *BMC Ophthalmol*. 2014 Apr 16;14(1):52.
63. 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. Absence of filaggrin mutation in a patient affected by pachyonychia congenita and mild atopic dermatitis. *Eur J Dermatol*. 2014 Nov-Dec;24(6):703-4.
64. Cascella R, Ragazzo M, Strafella C, Missiroli F, Borgiani P, Angelucci F, Marsella LT, Cusumano A, Novelli G, Ricci F, Giardina E. Age-related macular degeneration: insights into inflammatory genes. *J Ophthalmol*. 2014;2014:582842.
65. Robino C, Ralf A, Pasino S, De Marchi MR, Ballantyne KN, Barbaro A, Bini C, Carnevali E, Casarino L, Di Gaetano C, Fabbri M, Ferri G, Giardina E, Gonzalez A, Matullo G, Nutini AL, Onofri V, Piccinini A, Pigionica M, Ponzano E, Previderè C, Resta N, Scarnicci F, Seidita G, Sorçaburu-Cigliero S, Turrina S, Verzeletti A, Kayser M. Development of an Italian RM Y-STR haplotype database: Results of the 2013 GEFI collaborative exercise. *Forensic Sci Int Genet*. 2015 Mar;15:56-63.
66. Cascella R, Strafella C, Ragazzo M, Zampatti S, Borgiani P, Gambardella S, Pirazzoli A, Novelli G, Giardina E. Direct PCR: a new pharmacogenetic approach for the inexpensive testing of HLA-B*57:01. *Pharmacogenomics J*. 2015. Apr;15(2):196-200.
67. Fattorini P, Previderè C, Sorçaburu-Cigliero S, Marrubini G, Alù M, Barbaro AM, Carnevali E, Carracedo A, Casarino L, Consoloni L, Corato S, Domenici R, Fabbri M, Giardina E, Grignani P, Baldassarra SL, Moratti M, Nicolin V, Pelotti S, Piccinini A, Pitacco P, Plizza L, Resta N, Ricci U, Robino C, Salvaderi L, Scarnicci F, Schneider PM, Seidita G, Trizzino L, Turchi C, Turrina S, Vatta P, Vecchiotti C, Verzeletti A, De Stefano F. The molecular characterization of a depurinated trial DNA sample can be a model to understand the reliability of the results in forensic genetics. *Electrophoresis*. 2014 Nov;35(21-22):3134-44.
68. Cordiali-Fei P, Latini A, Trento E, Zampatti S, Ferraresi V, Cota C, Volpi S, D'agosto G, Bordignon V, Giardina E, Di Carlo A, Cristaudo A, Ensoli F. Familial Kaposi's Sarcoma in HHV8 infected subjects presenting the G-174C allele of the IL-6 promoter: a possible role for EBV? *Eur J Dermatol*. 2014 Jul-Aug;24(4):503-4.
69. Bowes J, Budu-Aggrey A, Huffmeier U, Uebe S, Steel K, Hebert HL, Wallace C, Massey J, Bruce IN, Bluett J, Feletar M, Morgan AW, Marzo-Ortega H, Donohoe G, Morris DW, Helliwell P,

- Ryan AW, Kane D, Warren RB, Korendowych E, Alenius GM, Giardina E, Packham J, McManus R, FitzGerald O, McHugh N, Brown MA, Ho P, Behrens F, Burkhardt H, Reis A, Barton A. Corrigendum: Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. *Nat Commun.* 2015 Jul 6;6:7741.
70. Bowes J, Loehr S, Budu-Aggrey A, Uebe S, Bruce IN, Feletar M, Marzo-Ortega H, Helliwell P, Ryan AW, Kane D, Korendowych E, Alenius GM, Giardina E, Packham J, McManus R, FitzGerald O, Brown MA, Behrens F, Burkhardt H, McHugh N, Huffmeier U, Ho P, Reis A, Barton A. PTPN22 is associated with susceptibility to psoriatic arthritis but not psoriasis: evidence for a further PsA-specific risk locus. *Ann Rheum Dis.* 2015 Apr 28.
 71. 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.
 72. Bowes J, Budu-Aggrey A, Huffmeier U, Uebe S, Steel K, Hebert HL, Wallace C, Massey J, Bruce IN, Bluett J, Feletar M, Morgan AW, Marzo-Ortega H, Donohoe G, Morris DW, Helliwell P, Ryan AW, Kane D, Warren RB, Korendowych E, Alenius GM, Giardina E, Packham J, McManus R, FitzGerald O, McHugh N, Brown MA, Ho P, Behrens F, Burkhardt H, Reis A, Barton A. Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. *Nat Commun.* 2015 Feb 5;6:6046.
 73. Bowes J, Budu-Aggrey A, Huffmeier U, Uebe S, Steel K, Hebert HL, Wallace C, Massey J, Bruce IN, Bluett J, Feletar M, Morgan AW, Marzo-Ortega H, Donohoe G, Morris DW, Helliwell P, Ryan AW, Kane D, Warren RB, Korendowych E, Alenius GM, Giardina E, Packham J, McManus R, FitzGerald O, McHugh N, Brown MA, Ho P, Behrens F, Burkhardt H, Reis A, Barton A. Corrigendum: Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. *Nat Commun.* 2015 Jul 6;6:7741.
 74. 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. Comparative analysis between saliva and buccal swabs as source of DNA: lesson from HLA-B*57:01 testing. 2015;16(10):1039-46. doi: 10.2217/pgs.15.59. Epub 2015 Jul 31.
 75. 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. FLG (filaggrin) null mutations and sunlight exposure: Evidence of a correlation. *J Am Acad Dermatol.* 2015 Sep;73(3):528-9. doi: 10.1016/j.jaad.2015.06.022.
 76. Cascella R, Strafella C, Germani C, Novelli G, Ricci F, Zampatti S, Giardina E. The Genetics and the Genomics of Primary Congenital Glaucoma. *Biomed Res Int.* 2015;2015:321291. doi: 10.1155/2015/321291. Epub 2015 Sep 16. Review
 77. Spitalieri P, Talarico RV, Botta A, Murdocca M, D'Apice MR, Orlandi A, Giardina E, Santoro M, Brancati F, Novelli G, Sangiuolo F. Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. *Cell Reprogram.* 2015 Aug;17(4):275-87.
 78. Rufini S, Ciccacci C, Politi C, Giardina E, Novelli G, Borgiani P. Stevens-Johnson syndrome and toxic epidermal necrolysis: an update on pharmacogenetics studies in drug-induced severe skin reaction. *Pharmacogenomics.* 2015 Nov;16(17):1989-2002. doi: 10.2217/pgs.15.128. Epub 2015 Nov 10. Review.
 79. Ferese R, Modugno N, Campopiano R, Santilli M, Zampatti S, Giardina E, Nardone A, Postorivo D, Fornai F, Novelli G, Romoli E, Ruggieri S, Gambardella S. Four Copies of SNCA Responsible for Autosomal Dominant Parkinson's Disease in Two Italian Siblings. *Parkinsons Dis.* 2015;2015:546462. doi: 10.1155/2015/546462. Epub 2015 Nov 9.
 80. Cascella R, Strafella C, Gambardella S, Longo G, Borgiani P, Sangiuolo F, Novelli G, Giardina E. Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. *Electrophoresis.* 2016 Mar;37(5-6):860-4. doi: 10.1002/elps.201500346. Epub 2016 Jan 15.

81. Pantic B, Borgia D, Giunco S, Malena A, Kiyono T, Salvatori S, De Rossi A, Giardina E, Sangiuolo F, Pegoraro E, Vergani L, Botta A. Reliable and versatile immortal muscle cell models from healthy and myotonic dystrophy type 1 primary human myoblasts. *Exp Cell Res*. 2016 Mar 1;342(1):39-51. doi: 10.1016/j.yexcr.2016.02.013. Epub 2016 Feb 19.
82. Mango R, Luchetti A, Sangiuolo R, Ferradini V, Briglia N, Giardina E, Ferrè F, Helmer Citterich M, Romeo F, Novelli G, Sangiuolo F. Next Generation Sequencing and Linkage Analysis for the Molecular Diagnosis of a Novel Overlapping Syndrome Characterized by Hypertrophic Cardiomyopathy and Typical Electrical Instability of Brugada Syndrome. *Circ J*. 2016;80(4):938-49. doi: 10.1253/circj.CJ-15-0685. Epub 2016 Mar 9.
83. Budu-Aggrey A, Bowes J, Loehr S, Uebe S, Zervou MI, Helliwell P, Ryan AW, Kane D, Korendowych E, Giardina E, Packham J, McManus R, FitzGerald O, McHugh N, Behrens F, Burkhardt H, Huffmeier U, Ho P, Martin J, Castañeda S, Goulielmos G, Reis A, Barton A. Replication of a distinct psoriatic arthritis risk variant at the IL23R locus. *Ann Rheum Dis*. 2016 Jul;75(7):1417-8. doi: 10.1136/annrheumdis-2016-209290. Epub 2016 Mar 25.
84. 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. Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. *Hum Genomics*. 2016 Apr 4;10:9
85. Ferese R, Zampatti S, Griguoli AM, Fornai F, Giardina E, Barrano G, Albano V, Campopiano R, Scala S, Novelli G, Gambardella S. A New Splicing Mutation in the L1CAM Gene Responsible for X-Linked Hydrocephalus (HSAS). *J Mol Neurosci*. 2016 Jul;59(3):376-81. doi: 10.1007/s12031-016-0754-3. Epub 2016 May 20.
86. Cascella R, Strafella C, Longo G, Maccarone M, Borgiani P, Sangiuolo F, Novelli G, Giardina E. Pharmacogenomics of multifactorial diseases: a focus on psoriatic arthritis. *Pharmacogenomics*. 2016 Jun;17(8):943-51. doi: 10.2217/pgs.16.20. Epub 2016 Jun 7.
87. 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. Biomolecular index of therapeutic efficacy in psoriasis treated by anti-TNF alpha agents. *G Ital Dermatol Venereol*. 2016 Sep 14.
88. Carracedo A, Giardina E, Mosquera-Miguel A, Manzo L, Alvarez-Iglesias V, Schneider PM. Making progress in education: The EUROFORGEN master degree pilot project in forensic genetics. *Forensic Sci Int Genet*. 2017 May;28:e12-e13. doi: 10.1016/j.fsigen.2017.03.006. Epub 2017 Mar 6.

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