



INFORMAZIONI PERSONALI

Nome

GIARDINA EMILIANO

Indirizzo

Università degli Studi di Roma "Tor Vergata"- facoltà di
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Dipartimento di Biomedicina e Prevenzione Laboratorio di
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Titolo attuale

Professore Associato in Genetica Medica

ESPERIENZA LAVORATIVA

Data (2001- 2022)

- Coordina con il Prof. Carlo Caltagirone la Piattaforma di Genomica della Rete degli Istituti IRCCS di Neuroscienze istituita dal Ministero della Salute
- Direttore del Laboratorio di Genetica Forense dell'Università degli Studi di Roma "Tor Vergata"
- Direttore del Laboratorio di Medicina Genomica della UILDM (Unione Italiana Lotta alla Distrofia Muscolare) sito presso l'Istituto Fondazione Santa Lucia di Roma
- Membro titolare del tavolo permanente interforze ed interdisciplinare a supporto della Banca dati Nazionale del DNA
- Responsabile per la Biologia Forense presso l'Ordine Nazionale dei Biologi
- Coordinatore nazionale del Gruppo di Lavoro per la Genetica Forense istituito dalla SIGU (Società Italiana di Genetica Forense)
- Direttore del Master di secondo livello in Genetica Forense attivato presso l'Università di Roma "Tor Vergata"
- Responsabile delle attività di ricerca nell'ambito di un finanziamento Europeo finalizzato alla realizzazione di sistemi innovativi di identificazione personale e

ATTIVITÀ SCIENTIFICA

tipizzazione di tracce biologiche ad uso forense

- Componente del nucleo di ricerca afferente al "Centro di Eccellenza per lo Studio del Rischio Genomico in Patologie Complesse Multifattoriali" istituito dal MIUR (Ministero dell'Istruzione, dell'Università e della Ricerca) presso la facoltà di Medicina e Chirurgia dell'Università "Tor Vergata" di Roma;
- Partecipa al Consorzio Internazionale per lo studio della suscettibilità genetica alla psoriasi;

ATTIVITÀ DIDATTICA

- Insegna Genetica Medica per il corso di Laurea Specialistica di Biotecnologie Mediche presso l'Università degli studi di Roma "Tor Vergata"
- Insegna Genetica Medica II per il corso di Laurea Specialistica di Biologia ed Evoluzione Umana presso l'Università degli studi di Roma "Tor Vergata"
- Insegna Genetica Medica per il corso di Laurea in Biotecnologie istituito dalla Facoltà di Scienze Matematiche Fisiche e Naturali di Urbino
- Insegna Genetica Oculare per la scuola di Specializzazione di Oftalmologia istituita presso l'Università degli studi di Roma "Tor Vergata"
- Insegna Genetica Medica per la scuola di Specializzazione di Nefrologia istituita presso l'Università degli studi di Roma "Tor Vergata"
- E' docente del Master Universitario di I livello in Biotecnologie istituito dalla facoltà di Scienze Matematiche Fisiche e Naturali dell'Università di Urbino "CarloBo"
- Insegna genetica medica nel dottorato di Immunologia e Biotecnologie Applicate istituito presso l'Università degli studi di Roma "Tor Vergata"
- E' direttore del Master di secondo livello in genetica forense, istituito presso l'Università degli Studi di Roma "Tor Vergata"
- Insegna genetica forense nel Master Universitario di II livello in Genetica Forense istituito presso l'Università degli studi di Roma "Tor Vergata"
- E' docente del dottorato di Tecnologie Avanzate in Biomedicina istituito presso l'Università degli studi di Roma "Tor Vergata"
- E' responsabile di una rubrica permanente di biologia forense sulla rivista ufficiale dell'Ordine Nazionale dei Biologi
- Organizza costantemente eventi di divulgazione scientifica nell'ambito della genetica forense e della diagnosi prenatale
- E' autore di un libro di testo ad uso universitario "Genetica Medica Pratica" (ed. Aracne)
- E' autore di diversi capitoli libri

ATTIVITÀ ASSISTENZIALE

- E' direttore del Laboratorio di Medicina Genomica della UILDM
- Effettua: diagnosi molecolare di patologie mendeliane in epoca pre e post-natale

**ISTRUZIONE E
FORMAZIONE**

Date (2000 – 2019)

2016: chiamato dalla facoltà di Medicina e Chirurgia per il ruolo di Professore Associato per il settore scientifico disciplinare MED/03 Genetica Medica.
2014: consegue l'abilitazione scientifica nazionale per la fascia di professore associato per il settore MED/03 Genetica Medica.
2013: E' designato quale membro titolare del tavolo permanente interforze ed interdisciplinare a supporto della Banca dati Nazionale del DNA.
2013: E' direttore del Laboratorio di Genetica Molecolare della UILDM (Unione Italiana Lotta alla Distrofia Muscolare) sito presso l'Istituto Fondazione Santa Lucia di Roma.
2013: E' responsabile del Laboratorio di Genetica Forense dell'Università degli Studi di Roma "Tor Vergata".
2013: E' direttore e fondatore della Scuola Permanente di Biologia Forense istituita presso l'Università degli Studi di Roma "Tor Vergata" e finanziata dall'Ordine Nazionale dei Biologi.
2012: E' responsabile per la Biologia Forense presso l'Ordine Nazionale dei Biologi. 2012: E' coordinatore nazionale del Gruppo di Lavoro per la Genetica Forense istituito dalla SIGU (Società Italiana di Genetica Forense).
2011: co-direttore del Master di secondo livello di Genetica Forense attivato presso l'Università di Roma "Tor Vergata".
2010: Diploma di Specializzazione in genetica medica presso la scuola di specializzazione di genetica medica dell'università di Roma Tor Vergata.
2009: riceve il premio "cavalierato giovanile" quale giovane di talento per i contributi scientifici offerti nel campo della genetica medica.
2006: consegue il titolo di dottore di ricerca in fisiopatologia della morte cellulare. 2004: co-responsabile dell'attività di unità di ricerca nell'ambito di un finanziamento Europeo finalizzato alla realizzazione di sistemi innovativi di identificazione personale e tipizzazione di tracce biologiche ad uso forense.
2004: risulta vincitore di concorso per 1 posto di ricercatore universitario per il settore scientifico disciplinare MED/03
2001-oggi: responsabile delle analisi di genetica forense svolte dalla sezione di Genetica dell'Università degli Studi di Roma "Tor Vergata".
2000: consegue il diploma di Laurea in Scienze Biologiche con la votazione di 110 su 110 e lode discutendo una tesi sperimentale dal titolo "Analisi molecolare di una forma dominante di gozzo multinodulare", relatore Prof.ssa Caterina Tanzarella; correlatore il professor Giuseppe Novelli.

- Nome e tipo di istituto di istruzione o formazione
- Principali materie / abilità professionali oggetto dello studio

Università degli studi di Roma "Tor Vergata" e "Roma Tre"

Medicina Genomica, Genetica medica, Diagnosi Prenatale, Genetica forense

**CAPACITÀ E COMPETENZE
PERSONALI**

MADRELINGUA

ITALIANO

ALTRE LINGUA

- Capacità di lettura
- Capacità di scrittura
 - Capacità di espressione orale

INGLESE

OTTIMO

OTTIMO

OTTIMO

CAPACITÀ E COMPETENZE
RELAZIONALI

OTTIME CAPACITÀ RELAZIONALI ACQUISITE DURANTE IL PERCORSO DI FORMAZIONE

CAPACITÀ E COMPETENZE
ORGANIZZATIVE

OTTIME CAPACITÀ ORGANIZZATIVE ACQUISITE GRAZIE ALLA GESTIONE DI PERSONE, PROGETTI E BILANCI

CAPACITÀ E COMPETENZE
TECNICHE

OTTIME CAPACITÀ NELL'UTILIZZO DI COMPUTER ED ATTREZZATURE SPECIFICHE. CONOSCENZA DI TUTTI I PROTOCOLLI APPLICATI ED UTILIZZATI DURANTE IL PERCORSO LAVORATIVO

Parametri bibliometrici (google scholar): Totale numero di pubblicazioni: >100

H index: 35

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PUBBLICAZIONI SU RIVISTE INTERNAZIONALI

1. Capon F, et al., Mapping a dominant form of multinodular goiter to chromosome Xp22. *Am J Hum Genet.* 2000 Oct;67(4):1004-7.
2. Giardina E, et al., Mutational analysis of Peroxiredoxin IV: exclusion of a positional candidate for multinodular goitre. *BMC Medical Genetics* 2002,23 July, 3:5.
3. Semprini S, et al., 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, et al., 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, et al., Role of genetics in prevention of coronary atherosclerosis. *Curr Opin Cardiol.* 2003 Sep;18(5):368-371.
6. Mango R, et al., 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, et al., (2003). Insight into genetics of atopic dermatitis: Future approaches and directions. *Journal of Investigative Dermatology*, 121(5), 1265-1265.
8. Giardina E, et al., 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. Giardina E, et al., Psoriatic Arthritis and CARD15 Gene Polymorphisms: No Evidence for Association in the Italian Population. *J Invest Dermatol* 2004 May;122(5):1106-7
10. Giardina E, et al., 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, et al., 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, et al., 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, et al., Transmission ratio distortion in the spinal muscular atrophy locus: data from 314 prenatal tests. *Neurology*, 2005; 65(10):1631-1635.
17. Concolino P, et al., 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, et al., PSORS2 markers are not associated with psoriatic arthritis in the Italian population. *Hum Hered*, 2006; 61(2):120-122.
19. Giardina E, et al., 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, et al., Gonadal mosaicism in hereditary angioedema. *Clin Genet*, 2006; 70(1):83-85.
21. Porzio O, et al., 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, et al., *Front Biosci*, 2007; 12:1563-1573.
23. Capoluongo E, et al., Mannose-binding lectin polymorphisms and pulmonary outcome in premature neonates: a pilot study. *Intensive Care Med.* 2007; 33(10):1787-94.
24. Giardina E, et al., 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, et al., 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, et al., Frequency assessment of SNPs for forensic identification in different populations. *FSIGEN* 2007 1(3-4):e1-3.
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 41. Ulrike Hüffmeier, et al., Missense variant in TRAF3IP2 associates with psoriatic arthritis and psoriasis. *Nat Genet.*
 42. Amy Strange, et al., Identification of novel psoriasis susceptibility loci and genetic interaction between HLA-C and ERAP1 provides evidence for an integrated pathogenic pathway *Nat Genet.*
 43. Riveira-Munoz E, He SM, et al., 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.
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- exercise. *Forensic Sci Int Genet.* 2015Mar;15:56-63.
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 76. Cascella R, et al., FLG (filaggrin) null mutations and sunlight exposure: Evidence of a correlation. *J Am Acad Dermatol.* 2015 Sep;73(3):528-9.
 77. Cascella R, et al., Comparative analysis between saliva and buccal swabs as source of DNA: lesson from HLA-B*57:01 testing. *Pharmacogenomics.* 2015;16(10):1039-46.
 78. Cascella R, et al., Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. *Electrophoresis.* 2015 Dec 17.
 79. Ferese R, et al., Four Copies of SNCA Responsible for Autosomal Dominant Parkinson's Disease in Two Italian Siblings. *Parkinsons Dis.* 2015;2015:546462.
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 81. Mango R, et al., 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:
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 86. Bianchi L, et al., Biomolecular index of therapeutic efficacy in psoriasis treated with anti-TNF- α agents. *G Ital Dermatol Venereol.* 2018 Jun;153(3):316-325.
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 90. Cascella R, et al., Towards the application of precision medicine in Age-Related Macular Degeneration. *Prog Retin Eye Res.* 2018 Mar;63:132-146.
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