

# GIOVANNI MALERBA

Data di nascita: 11 agosto 1971  
Luogo di nascita: Verona, Italia.  
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## Studi

	1990	Diploma di Liceo Scientifico, presso il Liceo <i>Piccole Figlie di S.Giuseppe</i> a Verona
	01/03/1996	Laurea in Scienze Biologiche (Marzo 1996), presso l' <i>Università degli Studi di Padova</i> , con una tesi dal titolo "Analisi di mutazioni in geni candidati in alcune patologie respiratorie: esame dei geni della Fibrosi Cistica, dell'alfa1-antitripsina e dell'alfa1-antichimotripsina"
15/10/2000	15/10/2000	Diploma di Specializzazione in Genetica Medica - indirizzo tecnico, presso l' <i>Università degli Studi di Verona</i> con una tesi dal titolo "Studio di linkage del gene NOS2 e del cromosoma 19 in asma allergico e utilizzo della simulazione per stimare il potere dell'analisi statistica"
	15/05/2006	Dottorato in Biotecnologie Applicate alle Scienze Biomediche presso l' <i>Università degli Studi di Verona</i> con una tesi dal titolo "Scansione genomica per asma allergico in un campione di famiglie asmatiche Italiane"

## Abilitazioni

	1997	Abilitazione all'esercizio della Professione di Biologo conseguita nella prima sessione dell'anno 1997
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## Posizioni Lavorative

1996	2003	Biologo Collaboratore, presso la Sezione di Biologia e Genetica del Dipartimento Materno Infantile di Biologia e Genetica dell' <i>Università degli Studi di Verona</i> (Prof. Pier Franco Pignatti). Durante questo periodo Giovanni è risultato vincitore di borse di studio per studi di linkage ed associazione del fattore genetico con caratteri complessi e ha conseguito la Specializzazione in Genetica - Indirizzo Tecnico.
2003 (aprile)	2006 (febbraio)	Posizione a tempo determinato di Tecnico/Amministrativo livello E durante la quale si sono sviluppati ed implementati metodi di biologia computazionale e statistica per lo studio della variabilità fenotipica e genotipica in malattie Mendeliane e malattie complesse.
2006 (marzo)	2014 (agosto)	Ricercatore Universitario(SSD: MED/03, Genetica Medica) - Dipartimento Materno Infantile e di Biologia e Genetica, <i>Università degli Studi di Verona</i> .
2014 (1 settembre)	oggi	Professore Associato (SSD: MED/03, Genetica Medica) - Dipartimento di Neuroscienze, Biomedicina e Movimento, <i>Università degli Studi di Verona</i> .

## Attività di ricerca presso istituti di ricerca esteri

15/10/1997	14/02/1998	Fox Chase Cancer Center (Filadelfia, USA) per studi di linkage a livello genomico in famiglie con coppie di fratelli affetti da asma allergica.
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10/01/2008	20/04/2008	Stephan Weiland Fellowship ( Progetto EU Gabriel ), per lo studio intitolato "Pooled analysis of genome-wide linkage scans from 3 European family samples" ( Prof. F. Demenais, Fondation Jean Dausset-Centre d'Etude du Polymorphisme Humain - CEPH- , Paris, FranceParigi)
05/05/2009	30/06/2009	INSERM U794, CEPH institute, Paris (Prof. Florence Demenais) per uno studio di meta-analisi di studi linkage per l'asma allergica in famiglie europee
05/10/2009	30/03/2010	INSERM U794, CEPH institute, Paris (Prof. Florence Demenais). Fellowship ERS/AIMAR per uno studio sugli effetti dell'imprinting nell'asma allergica basati su analisi di linkage e di associazione a livello genomico.
15/09/2010	20/10/2010	Sanger Center, Hinxton, UK (Prof.ssa Nicole Soranzo) per uno studio sul controllo di qualità delle genotipizzazioni ottenute tramite chip array in un campione di 1000 individui Italiani e successiva imputazione dei genotipi tramite il pannello di riferimento del progetto HAPMAP2
02/10/2010	30/10/2010	Sanger Center, Hinxton, UK (Prof.ssa Nicole Soranzo) per uno studio sul profilo lipidico degli individui della coorte INCIPE (4000 individui) ed imputazione di tutti gli individui genotipizzati utilizzando i pannelli del progetto 1000 genomi e dello studio UK10K

## Riassunto Attività Scientifica

Ho iniziato l'attività scientifica da studente universitario in Biologia studiando la componente genetica delle malattie multifattoriali respiratorie (settembre 1994 - marzo 1996). Durante i successivi anni, da specializzando in Genetica Medica (indirizzo tecnico), ho approfondito le conoscenze sui metodi statistici per caratterizzare e studiare la componente genetica sia nelle malattie Mendeliane che nelle malattie complesse.

Nel 1997 mi sono unito al gruppo di bioinformatica del Dr. KH. Buetow, un esperto nella comunità della ricerca biomedica per l'integrazione e l'analisi dei dati genetici e la loro applicazione all'assistenza del paziente, al Fox Chase Cancer Center (Filadelfia, USA) per 4 mesi (ottobre 1997 - febbraio 1998). Durante quel periodo ho avuto modo di imparare a programmare con diversi linguaggi di programmazione al calcolatore e di comprendere in la logica dei metodi statistici applicati alle analisi di linkage a livello genomico. Durante il periodo della specializzazione ho avuto modo di accrescere le mie conoscenze di informatica (programmazione in ambito bioinformatico con diversi linguaggi, gestione e amministrazione di database e di applicazioni web) combinandole alle conoscenze di genetica medica. Nello stesso periodo ho migliorato le conoscenze nell'ambito della genomica computazionale e della biostatistica. Queste conoscenze mi hanno poi permesso di poter collaborare con gruppi di ricerca sia dell'Università di Verona che di altre Università Italiane.

Dal gennaio ad aprile 2008, mi sono unito al gruppo di ricerca della Prof.ssa Demenais a Parigi (INSERM U794, Istituto CEPH) dove ho sviluppato le mie conoscenze nell'epidemiologia genetica mediante l'applicazione di diversi approcci statistici per studiare il linkage e l'associazione con fenotipi complessi in studi familiari. Ho potuto poi riunirmi al gruppo della prof.ssa Demenais dal novembre 2009 al marzo 2010 per definire ed implementare modelli statistici adatti a studiare trasmissioni alleliche preferenziali da parte di un solo genitore nell'asma infantile.

Dal settembre 2010 ad ottobre 2010, mi sono unito al gruppo della Dr. Nicole Soranzo del Sanger Center (Hinxton, Regno Unito) dove ho lavorato sui dati genetici e fenotipici per la realizzazione di una scansione genomica (milioni di marcatori del DNA) in un campione di circa 1000 individui Italiani (coorte dello studio INCIPE, rivolto allo studio di diversi aspetti della malattia renale - referenti: prof Lupu, prof Gambaro). Al Sanger Center ho imparato ad utilizzare sofisticati strumenti bioinformatici e statistici per realizzare i controlli di qualità sul dato che precedono lo studio di associazione ed a eseguire l'imputazione dei genotipi a partire da un pannello di individui ben caratterizzati a livello genomico (i campioni del progetto Hapmap prima e i campioni progetto 1000genomi poi). Durante questa esperienza ho imparato ad utilizzare i cluster di computer dedicati al calcolo ad alte prestazioni.

A partire dal 2009 ho avuto modo di lavorare sui dati prodotti dalle recenti piattaforme dedicate al sequenziamento di nuova generazione (NGS). Trattandosi di una tecnologia nuova, abbiamo dovuto sviluppare gli strumenti bioinformatici e statistici per poter lavorare sui dati ottenuti. L'interesse è stato inizialmente rivolto a studi del trascrittoma umano (malattie dello spettro autistico, Alzheimer, malattia coronarica, linfomi, leucemie, trapiantati di rene) e anche di alcune piante. Nel 2010 sfruttando quanto appreso durante la mia precedente permanenza al Sanger Center mi sono unito ai consorzi internazionali CKDGen e GUGC nella qualità di ricercatore coinvolto nella analisi statistica dei dati per lo studio della componente genetica dei fenotipi associati alla funzionalità renale. Tali consorzi includono molti gruppi internazionali che contribuiscono con la loro casistica di individui caratterizzati a livello genomico e ben fenotipizzati per i caratteri di interesse a realizzare meta-analisi di centinaia di migliaia di individui. A partire dal 2010 mi sono cointeressato dell'analisi del dato genetico prodotto da piattaforme di NGS. Nel tempo ho avuto modo di condurre studi su coorti di esomi, di trascrittomi e più recentemente di metilomi, sempre nel contesto di uno studio di linkage o associazione del fattore genetico con un fenotipo complesso.

### **Revisore di articoli per riviste scientifiche**

2005            oggi  
Allergy, American Journal of Case Reports, American Journal of Respiratory and Critical Care Medicine, American Journal of Respiratory and Critical Care, Clinical Chemistry, Experimental Biology and Medicine, Gene, Human Genetics, Journal of Human Genetics, Journal of Nutrition and Metabolism, Nutrition & Metabolism, Plos One, Respiratory Research e The Pharmacogenomics Journal

### **Capitoli di libri**

Capitolo 17. "Tecniche Genomiche, bioinformtica e statistica abbinata alla genetica" (Pp 369-397) all'interno del libro Nutrigenomica ed epigenetica. Dalla biologia alla clinica di Damiano Galimberti, Giovanni Battista Gidaro, Vittorio Calabrese.  
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# Elenco delle pubblicazioni (aggiornato al 12 giugno 2018)

## Articoli pubblicati su riviste internazionali peer-reviewed:

H-index (Scopus): 34

Numero di citazioni (Scopus): 3702

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Numero articolo presenti in JCR (2006): 92

1. Mahajan A, Wessel J, Willems SM, Zhao W, Robertson NR, Chu AY, Gan W, Kitajima H, Taliun D, Rayner NW, Guo X, Lu Y, Li M, Jensen RA, Hu Y, Huo S, Lohman KK, Zhang W, Cook JP, Prins BP, Flannick J, Grarup N, Trubetskoy VV, Kravic J, Kim YJ, Rybin DV, Yaghootkar H, Müller-Nurasyid M, Meidtner K, Li-Gao R, Varga TV, Marten J, Li J, Smith AV, An P, Ligthart S, Gustafsson S, **Malerba G**, Demirkan A, Tajes JF, Steinthorsdottir V, Wuttke M, Lecoeur C, Preuss M, Bielak LF, Graff M, Highland HM, Justice AE, Liu DJ, Marouli E, Peloso GM, Warren HR; ExomeBP Consortium; MAGIC Consortium; GIANT Consortium; Afaq S, Afzal S, Ahlqvist E, Almgren P, Amin N, Bang LB, Bertoni AG, Bombieri C, Bork-Jensen J, Brandslund I, Brody JA, Burt NP, Canoui M, Chen YI, Cho YS, Christensen C, Eastwood SV, Eckardt KU, Fischer K, Gambaro G, Giedraitis V, Grove ML, de Haan HG, Hackinger S, Hai Y, Han S, Tybjaerg-Hansen A, Hivert MF, Isomaa B, Jäger S, Jørgensen ME, Jørgensen T, Käräjämäki A, Kim BJ, Kim SS, Koistinen HA, Kovacs P, Kriebel J, Kronenberg F, Läll K, Lange LA, Lee JJ, Lehne B, Li H, Lin KH, Linneberg A, Liu CT, Liu J, Loh M, Mägi R, Mamakou V, McKean-Cowdin R, Nadkarni G, Neville M, Nielsen SF, Ntalla I, Peyser PA, Rathmann W, Rice K, Rich SS, Rode L, Rolandsson O, Schönherr S, Selvin E, Small KS, Stančáková A, Surendran P, Taylor KD, Teslovich TM, Thorand B, Thorleifsson G, Tin A, Tönjes A, Varbo A, Witte DR, Wood AR, Yajnik P, Yao J, Yengo L, Young R, Amouyel P, Boeing H, Boerwinkle E, Bottinger EP, Chowdhury R, Collins FS, Dedoussis G, Dehghan A, Deloukas P, Ferrario MM, Ferrières J, Florez JC, Frossard P, Gudnason V, Harris TB, Heckbert SR, Howson JMM, Ingelsson M, Kathiresan S, Kee F, Kuusisto J, Langenberg C, Launer LJ, Lindgren CM, Männistö S, Meitinger T, Melander O, Mohlke KL, Moitry M, Morris AD, Murray AD, de Mutsert R, Orho-Melander M, Owen KR, Perola M, Peters A, Province MA, Rasheed A, Ridker PM, Rivadineira F, Rosendaal FR, Rosengren AH, Salomaa V, Sheu WH, Sladek R, Smith BH, Strauch K, Uitterlinden AG, Varma R, Willer CJ, Blüher M, Butterworth AS, Chambers JC, Chasman DI, Danesh J, van Duijn C, Dupuis J, Franco OH, Franks PW, Froguel P, Grallert H, Groop L, Han BG, Hansen T, Hattersley AT, Hayward C, Ingelsson E, Kardia SLR, Karpe F, Kooner JS, Köttgen A, Kuulasmaa K, Laakso M, Lin X, Lind L, Liu Y, Loos RJF, Marchini J, Metspalu A, Mook-Kanamori D, Nordestgaard BG, Palmer CNA, Pankow JS, Pedersen O, Psaty BM, Rauramaa R, Sattar N, Schulze MB, Soranzo N, Spector TD, Stefansson K, Stumvoll M, Thorsteinsdottir U, Tuomi T, Tuomilehto J, Wareham NJ, Wilson JG, Zeggini E, Scott RA, Barroso I, Frayling TM, Goodarzi MO, Meigs JB, Boehnke M, Saleheen D, Morris AP, Rotter JI, McCarthy MI. Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. *Nat Genet.* 2018 Apr;50(4):559-571. doi: 10.1038/s41588-018-0084-1. Epub 2018 Apr 9. PubMed PMID: 29632382; PubMed Central PMCID: PMC5898373. [IF: 27.959]
2. Andreini A, Zampieri N, Costantini C, **Malerba G**, Bruno C, Salvagno G, Francia G, Gottardi M, Rimondini M, Ferrarini I, Lippi G, Ambrosetti A, Benedetti F, Tecchio C. Chronic graft versus host disease is associated with erectile dysfunction in allogeneic hematopoietic stem cell transplant patients: a single-center experience. *Leuk Lymphoma.* 2018 Mar 21:1-4. doi: 10.1080/10428194.2018.1443338. [Epub ahead of print] PubMed PMID: 29561209. [IF: 2.755]
3. Cecconi D, Carbonare LD, Mori A, Cheri S, Deiana M, Brandi J, Degaetano V, Masiero V, Innamorati G, Mottes M, **Malerba G**, Valenti MT. An integrated approach identifies new oncotargets in melanoma. *Oncotarget.* 2017 Dec 15;9(14):11489-11502. doi: 10.18632/oncotarget.23727. eCollection 2018 Feb 20. PubMed PMID: 29545914; PubMed Central PMCID: PMC5837771. [IF: 5.168]
4. Kraja AT, Cook JP, Warren HR, Surendran P, Liu C, Evangelou E, Manning AK, Grarup N, Drenos F, Sim X, Smith AV, Amin N, Blakemore AIF, Bork-Jensen J, Brandslund I, Farmaki AE, Fava C, Ferreira T, Herzig KH, Giri A, Giulianini F, Grove ML, Guo X, Harris SE, Have CT, Havulinna AS, Zhang H, Jørgensen ME, Käräjämäki A, Kooperberg C, Linneberg A, Little L, Liu Y, Bonnycastle LL, Lu Y, Mägi R, Mahajan A, **Malerba G**, Marioni RE, Mei H, Menni C, Morrison AC, Padmanabhan S, Palmas W, Poveda A, Rauramaa R, Rayner NW, Riaz M, Rice K, Richard MA, Smith JA, Southam L, Stančáková A, Stirrups KE, Tragante V, Tuomi T, Tzoulaki I, Varga TV, Weiss S, Yiorkas AM, Young R, Zhang W, Barnes MR, Cabrera CP, Gao H, Boehnke M, Boerwinkle E, Chambers JC, Connell JM, Christensen CK, de Boer RA, Deary IJ, Dedoussis G, Deloukas P, Dominiczak AF, Dörr M, Joehanes R, Edwards TL, Esko T, Fornage M, Franceschini N, Franks PW, Gambaro G, Groop L, Hallmans G, Hansen T, Hayward C, Heikki O, Ingelsson E, Tuomilehto J, Jarvelin MR, Kardia SLR, Karpe F, Kooner JS, Lakka TA, Langenberg C, Lind L, Loos RJF, Laakso M, McCarthy MI, Melander O, Mohlke KL, Morris AP, Palmer CNA, Pedersen O, Polasek O, Poulter NR, Province MA, Psaty BM, Ridker PM, Rotter JI, Rudan I, Salomaa V, Samani NJ, Sever P, Skaaby T, Stafford JM, Starr JM, van der Harst P, van der Meer P; Understanding Society Scientific Group, van Duijn CM, Vergnaud AC, Gudnason V, Wareham NJ, Wilson JG, Willer CJ, Witte DR, Zeggini E, Saleheen D, Butterworth AS, Danesh J, Asselbergs FW, Wain LV, Ehret GB, Chasman DI, Caulfield MJ, Elliott P, Lindgren CM, Levy D, Newton-Cheh C, Munroe PB, Howson JMM; CHARGE EXOME BP, CHD Exome+, Exome BP, GoT2D:T2DGenes Consortia, The UK Biobank Cardio-Metabolic Traits Consortium Blood Pressure Working Group. New Blood Pressure-Associated Loci Identified in Meta-Analyses of 475 000 Individuals. *Circ Cardiovasc Genet.* 2017 Oct;10(5). pii: e001778. doi: 10.1161/CIRCGENETICS.117.001778. PubMed PMID: 29030403; PubMed Central PMCID: PMC5776077. [IF: 4.743]
5. Rosendahl J, Kirsten H, Hegyi E, Kovacs P, Weiss FU, Laumen H, Lichtner P, Ruffert C, Chen JM, Masson E, Beer S, Zimmer C, Seltsam K, Algül H, Bühler F, Bruno MJ, Bugert P, Burkhardt R, Cavestro GM, Cichoz-Lach H, Farré A, Frank J, Gambaro G, Gimpfl S, Grallert H, Griesmann H, Grützmann R, Hellerbrand C, Hegyi P, Hollenbach M,

- Iordache S, Jurkowska G, Keim V, Kiefer F, Krug S, Landt O, Leo MD, Lerch MM, Lévy P, Löffler M, Löhr M, Ludwig M, Macek M, Malats N, Malecka-Panas E, **Malerba G**, Mann K, Mayerle J, Mohr S, Te Morsche RHM, Motyka M, Mueller S, Müller T, Nöthen MM, Pedrazzoli S, Pereira SP, Peters A, Pfützner R, Real FX, Rebours V, Ridinger M, Rietschel M, Rösmann E, Saftoiu A, Schneider A, Schulz HU, Soranzo N, Soyka M, Simon P, Skipworth J, Stickel F, Strauch K, Stumvoll M, Testoni PA, Tönjes A, Werner L, Werner J, Wodarz N, Ziegler M, Masamune A, Mössner J, Férec C, Michl P, P H Drenth J, Witt H, Scholz M, Sahin-Tóth M; all members of the PanEuropean Working group on ACP. Genome-wide association study identifies inversion in the CTRB1-CTRB2 locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. *Gut*. 2017 Jul 28. pii: gutjnl-2017-314454. doi: 10.1136/gutjnl-2017-314454. [Epub ahead of print] PubMed PMID: 28754779. [IF: 16.658]
6. Sangalli A, Orlandi E, Poli A, Maurichi A, Santinami M, Nicolis M, Ferronato S, **Malerba G**, Rodolfo M, Gomez Lira M. Sex-specific effect of RNASEL rs486907 and miR-146a rs2910164 polymorphisms' interaction as a susceptibility factor for melanoma skin cancer. *Melanoma Res*. 2017 Aug;27(4):309-314. doi: 10.1097/CMR.0000000000000360. PubMed PMID: 28654546. [IF: 2.615]
  7. Dalle Carbonare L, Mottes M, **Malerba G**, Mori A, Zaninotto M, Plebani M, Dellantonio A, Valenti MT. Enhanced Osteogenic Differentiation in Zoledronate-Treated Osteoporotic Patients. *Int J Mol Sci*. 2017 Jun 13;18(6). pii: E1261. doi: 10.3390/ijms18061261. PubMed PMID: 28608802; PubMed Central PMCID: PMC5486083. [IF: 3.226]
  8. Tachmazidou I, Süveges D, Min JL, Ritchie GRS, Steinberg J, Walter K, Iotchkova V, Schwartzentruber J, Huang J, Memari Y, McCarthy S, Crawford AA, Bombieri C, Cocca M, Farmaki AE, Gaunt TR, Jousilahti P, Kooijman MN, Lehne B, **Malerba G**, Männistö S, Matchan A, Medina-Gomez C, Metrustry SJ, Nag A, Ntalla I, Paternoster L, Rayner NW, Sala C, Scott WR, Shihab HA, Southam L, St Pourcain B, Traglia M, Trajanoska K, Zaza G, Zhang W, Artigas MS, Bansal N, Benn M, Chen Z, Danecsek P, Lin WY, Locke A, Luan J, Manning AK, Mulas A, Sidore C, Tybjaerg-Hansen A, Varbo A, Zoledziewska M, Finan C, Hatzikotoulas K, Hendricks AE, Kemp JP, Moayyeri A, Panoutsopoulou K, Szpak M, Wilson SG, Boehnke M, Cucca F, Di Angelantonio E, Langenberg C, Lindgren C, McCarthy MI, Morris AP, Nordestgaard BG, Scott RA, Tobin MD, Wareham NJ; SpiroMeta Consortium; GoT2D Consortium, Burton P, Chambers JC, Smith GD, Dedoussis G, Felix JF, Franco OH, Gambaro G, Gasparini P, Hammond CJ, Hofman A, Jaddoe VVW, Kleber M, Kooner JS, Perola M, Relton C, Ring SM, Rivadeneira F, Salomaa V, Spector TD, Stegle O, Toniolo D, Uitterlinden AG; arcOGEN Consortium; Understanding Society Scientific Group; UK10K Consortium, Barroso I, Greenwood CMT, Perry JRB, Walker BR, Butterworth AS, Xue Y, Durbin R, Small KS, Soranzo N, Timpson NJ, Zeggini E. Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *Am J Hum Genet*. 2017 Jun 1;100(6):865-884. doi: 10.1016/j.ajhg.2017.04.014. Epub 2017 May 25. PubMed PMID: 28552196; PubMed Central PMCID: PMC5473732. [IF: 9.025]
  9. Mottes M, **Malerba G**. Hunting Novel Human Disease Genes in the Next Generation Sequencing Era: Lessons from Osteogenesis Imperfecta. *J Genet Genom*. 2017. 1(1):e102. [ultimo autore][rivista non recensita da jcr] [IF: non censito in JCR]
  10. De Summa S, **Malerba G**, Pinto R, Mori A, Mijatovic V, Tommasi S. GATK hard filtering: tunable parameters to improve variant calling for next generation sequencing targeted gene panel data. *BMC Bioinformatics*. 2017 Mar 23;18(Suppl 5):119. doi: 10.1186/s12859-017-1537-8. PubMed PMID: 28361668; PubMed Central PMCID: PMC5374681. [co-primo autore e autore corrispondente] [IF: 2.448]
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