





INFORMAZIONI PERSONALI

Prof. Giuseppe Matullo

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<https://www.iigm.it/site/>

Sesso M | Data di nascita | Nazionalità Italiana

POSIZIONE ATTUALE

- Professore Ordinario di Genetica Medica MED/03 presso il Dipartimento di Scienze Mediche, Scuola di Medicina, Università di Torino (dal 2018; Professore Associato dal 2006)
- Dirigente Ospedale Molinette, Servizio di Genetica Medica, Città della Salute e della Scienza, Torino (dal 2015)
- Direttore dell'Unità "Genomic Variation, Complex Diseases and Population Medicine" presso il Dipartimento di Scienze Mediche), Università di Torino (dal 2006)
- Responsabile della Piattaforma di Analisi Genomica presso il DSM (dal 2018)

AREE DI RICERCA

- Genetica delle malattie complesse e interazioni geni-ambiente
- Genetica medica e di popolazioni
- Suscettibilità genetica nelle malattie cardiovascolari e metaboliche
- Genetica ed epidemiologia molecolare dei tumori
- Correlazione genotipo-fenotipo in relazione alla capacità di riparare i danni del DNA
- Polimorfismi nei geni della riparazione del DNA, suscettibilità tumorale e risposta alla terapia
- Genomica delle popolazioni umane e studi di associazione genome-wide nei tumori e in patologie cardiovascolari e metaboliche
- Epigenomica delle malattie complesse e cambiamenti epigenetici dovuti a esposizioni ambientali, dieta e stile di vita
- Analisi di mutazioni e della variabilità genetica umana tramite Next Generation Sequencing (NGS) su piattaforme Illumina (HiScanSQ e MiSeq): pannelli custom ed exome sequencing (Agilent) e TruSight Cancer panel (Illumina).
- Whole-genome sequencing in campioni di popolazione ben caratterizzati dell'Italian Genome project

SINOSI DELLA RICERCA

Il prof. Giuseppe Matullo è ordinario di genetica umana presso il Dipartimento di Scienze Mediche, Università di Torino (Italia), Facoltà di Medicina e PI della Unità di Variazione Genomica e di Ricerca Traslazionale dell'Istituto Italiano di Medicina Genomica (IIGM, ex Human Genetics Foundation - HuGeF), Torino. È un esperto riconosciuto a livello internazionale nell'analisi di studi di associazione genome- ed epigenome-wide (GWAS ed EWAS) di tratti complessi, cancro (ad es. vescica e mesotelioma), malattie cardiovascolari (CVD) e altre malattie croniche. Le competenze del Prof. Matullo coprono diversi aspetti della genetica molecolare delle malattie complesse, dalla variazione genomica, alla trascrizione, alla regolazione dell'espressione (metilazione del DNA e miRNA), tutte da indagare congiuntamente con l'esposizione ambientale ed i fattori dello stile di vita (interazioni gene-ambiente). Ha anche studiato la lunghezza dei telomeri e la metilazione del DNA come misura dell'età biologica e come risultato dell'interazione tra fattori genetici e ambientali nello sviluppo di malattie croniche. Il gruppo del prof. Matullo ha una lunga esperienza nell'applicazione di tecniche di genetica molecolare ad alta processività (High Throughput) per lo studi di ampie popolazioni (ad esempio, lo studio EPIC, 500.000 volontari in tutta Europa). In particolare, il suo gruppo è stato coinvolto per molti anni nell'analisi della correlazione genotipo-fenotipo della riparazione del DNA, nell'identificazione di biomarcatori correlati all'alimentazione e al cancro / CVD (progetto ECNIS, EPIC-CVD). La lunghezza dei telomeri dei leucociti (LTL) è stata misurata in EPIC-CVD (42.000 individui) e in altre malattie croniche in diversi progetti collaborativi in relazione ad esposizioni / stile di vita ambientale, attività di riparazione del DNA, stress ossidativo, ecc. Negli ultimi anni ha utilizzato molti approcci di sequenziamento Next Generations che includono ampi pannelli genici, sequenziamento dell'esoma e dell'intero genoma. Il suo gruppo ha svolto numerose ricerche nel campo della medicina traslazionale in oncologia (tra i quali il cancro alla vescica e il mesotelioma) per l'identificazione di biomarcatori omici (in particolare la variazione genomica, la metilazione del DNA e il profilo dei

miRNA) che possano essere utilizzati per la diagnosi precoce, la stratificazione dei pazienti per la prognosi e terapie personalizzate. In particolare, ha ottenuto un brevetto basato su miRNA in grado di identificare, mediante un dosaggio urinario non invasivo, pazienti con carcinoma della vescica da controlli sani (brevetto UROMIRNA).

L'analisi dei profili di metilazione del DNA nelle cellule del sangue sta permettendo anche di ottenere informazioni specifiche sul reale invecchiamento biologico individuale, in particolare in relazione all'invecchiamento del sistema immunitario, con nuove intuizioni sul potenziale intervento di immunoprevenzione e immunoterapia.

Nel 2016 il Prof. Matullo ha fondato, insieme ad altri noti genetisti italiani e con il sostegno della Società Italiana di Genetica Umana (SIGU), il Network dei genomi Italiani (NIG; <http://www.nig.cineca.it/>); una partnership che include diverse università e ospedali pubblici (Torino, Milano, Pavia, Bologna, Trieste, Siena, Roma, Napoli) con l'obiettivo di raccogliere migliaia di esomi e genomi prodotti mediante sequenziamento massivo parallelo, al fine di avere un quadro completo della distribuzione di varianti genetiche / aplotipi in Italia per scopi di genetica clinica e di popolazione, applicazioni forensi e di farmacogenetica.

ESPERIENZE LAVORATIVE

- 2006-2018: Professore Associato di Genetica Medica MED/03 presso il Dipartimento di Scienze Mediche, Scuola di Medicina, Università di Torino
- 2010-2018: Direttore dell'Unità "Genomic Variation and Translational Research" presso l'Italian Institute for Genomic Medicine (IIGM – ex HuGeF), Torino
- 2006-2010: Vice-direttore della Sezione Life-Sciences presso la Fondazione I.S.I (Institute for Scientific Interchange), Torino
- 2000-2010: Responsabile dei laboratori di Genetica ed Epidemiologia Molecolare della Fondazione I.S.I (Institute for Scientific Interchange), Torino
- 1998-2006: Tecnico di ricerca presso il Dip. di Genetica, Biologia e Biochimica, Università di Torino, Facoltà di Medicina e Chirurgia
- Ago-Ott 1997: ricercatore borsista presso il Rangos Research Center (Università di Pittsburgh, PA, USA) nel laboratorio di Genetica Molecolare (Immunogenetica) diretto dal Prof. Massimo Trucco.
- Agosto 1991: ricercatore borsista presso il laboratorio del Prof. L.L. Cavalli-Sforza, Dip. di Genetica, Università di Stanford University, CA.
- Sett-Nov 1991: ricercatore borsista presso il Rangos Research Center (Università di Pittsburgh, PA, USA) nel laboratorio di Genetica Molecolare (Immunogenetica) diretto dal Prof. Massimo Trucco.

ISTRUZIONE E FORMAZIONE

- Ottobre 1999 **MSc**
 Università di Pavia, European Schools for Advanced Studies, Italia
 - Statistical Genetics
- Aprile 1997 **PhD**
 Università di Torino, Italia
 - Genetica Umana
- Agosto-Ottobre 1997
 - Ricercatore borsista presso il Rangos Research Center, Università di Pittsburgh (PA, USA), Prof. Massimo Trucco
- March 1991 **BScD**
 Università di Torino, Italia
 - Scienze Biologiche
- Agosto-Novembre 1991
 - Ricercatore Borsista, Dipartimento di Genetica, Università di Stanford (CA, USA), Prof. L.L. Cavalli-Sforza.
 - Ricercatore Borsista presso il Rangos Research Center, Università di Pittsburgh (PA, USA), Prof. Massimo Trucco

ATTIVITA' DI INSEGNAMENTO

- Dal 1998 Insegnamento di Genetica umana e medica nel corso di Tecnico di laboratorio, Tecnico di Riabilitazione, Fisioterapista, Ortopedico e Logopedista presso la Scuola di Medicina dell'Università di Torino, Italia.
- Dal 1998 Insegnamento di Genetica Medica nei corsi di specializzazione in Genetica e Cardiologia Medica,

2001 e 2003	Università di Torino, Italia. Organizzazione e insegnamento nel "Corso Internazionale di Epidemiologia Molecolare", I.S.I. Fondazione, Villa Gualino, Torino, Italia, quarta e sesta edizione.
2002-2010	Organizzazione e insegnamento nel corso di Epidemiologia molecolare presso il Dipartimento di Scienze Biomediche e Oncologia Umana dell'Università di Torino, Italia.
Dal 2003	Consiglio della Scuola di Dottorato in Scienze Biomediche e Oncologia Umana, Università di Torino, Italia.
Dal 2006	Insegnamento di Genetica umana e medica, Facoltà di Medicina e Scuola di Medicina, Università di Torino, Italia.
Dal 2011	Coordinatore del programma di genetica umana nell'ambito della scuola di dottorato in oncologia e scienze biomediche
Dal 2011	Insegnamento di genetica medica presso la Scuola di scienze alimentari e nutrizione umana, Università di Torino, Italia.
Dal 2017	Comitato didattico e di facoltà del Master in Immunogenetica e biologia dei trapianti
Dal 2017	Insegnamento al Master of Psico-Neuro-Endocrino-Immunologia (PNEI)

FINANZIAMENTI

- Progetti**
- Italian Association of Cancer Research (AIRC) grant (2004-2007): "EPIC-Italy: a molecular epidemiology prospective project on diet, genetic susceptibility and cancer risk".
 - Italian Association of Cancer Research (AIRC) grant (2004-2007): "Network of molecular epidemiology of cancer in Italy"
 - European Community grant "ECNIS" (2005-2010): ECNIS (Environmental Cancer Risk, Nutrition and Individual Susceptibility) is a Network of Excellence operating in the context of the 6th EU Framework Programme for Research and Development (FP6).
 - Italian Association of Cancer Research (AIRC) grant (2006-2008): "DNA repair genotype-phenotype correlation and cancer-risk". (P.I.)
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2007" (2007-2008), Direzione di Sanità Pubblica: "Identification of functional polymorphisms in DNA repair genes in lymphoblastoid cell lines: cancer risk and response to therapy". (P.I.)
 - MIUR ex60% (2007): "DNA repair capacity interindividual variability in relation to prognosis and chemosensitivity". (P.I.)
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2008" (2008-2009), Direzione di Sanità Pubblica: "Mutation detection in exfoliated bladder cells in urine for the diagnosis and the monitoring of bladder cancer". (P.I.)
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2008bis" (2008-2009), Direzione di Sanità Pubblica: "Identification of genes/polymorphisms involved in bladder cancer through replication of genome-wide association studies". (P.I.)
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2008bis" (2008-2009), Direzione di Sanità Pubblica: "Replication of genome-wide association studies on an independent sample of young AMI patients".
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2009" (2009-2010), Direzione di Sanità Pubblica: "Endometrial cancer and diet risk factors. A case-control study in the Turin population". (P.I.)
 - Grant "Progetti di ricerca sanitaria finalizzata anno 2009" (2009-2010), Direzione di Sanità Pubblica: "Association between leucocyte telomere length and AMI in young patients".
 - Grant Project EPICOR2 San Paolo (2008-2010): Risk of cardiovascular events associated to life style, biomarkers and genetic susceptibility in the frame of the EPICOR Italian and European collaboration. (P.I.)
 - European Community grant "ECNIS2" (2011-2013): ECNIS (Environmental Cancer Risk, Nutrition and Individual Susceptibility) (FP7).
 - Grant Medical Research Council (MRC) and the British Heart Foundation (BHF): "EPIC-Heart" (2010-2012), Cambridge, UK
 - European Community grant "EPIC-CVD 7th EU Framework Programme (FP7).
 - MIUR ex60% (2012): "Analisi epigenomica nell'infarto del miocardio in relazione a stile di vita, indicatori biologici e di suscettibilità genetica nell'ambito dello studio prospettico EPIC-Italia". (P.I.)
 - Grant Hufef to the Unit "Genomic Variation of Human Population and Complex Diseases (2010-2015)"
 - Grant Fondazione Veronesi (2013-2015): DNA REPAIR CAPACITY, TELOMERE LENGTH AND EPIGENETIC CHANGES AS AGING BIOMARKERS IN BLADDER CANCER. (P.I.)
 - Grant AIRC IG 17464 "Blood DNA methylation changes and plasma microRNA associated to asbestos-exposure and Malignant Pleural Mesothelioma" (2016-2018) (P.I.)
 - Grant supported by the Italian Ministry for Education and Research – MIUR project (2018 – 2022), "Departments of Excellence 2018 – 2022" TESEO Traguardi di Eccellenza nelle Scienze mediche Esplorando le Omiche.
 - Grant AIRC IG (2019-2023) Non-invasive predictive and prognostic biomarkers in Malignant Pleural Mesothelioma: from preclinical to clinical models (PI)

- Grant H2020-JTI-IMI2-2017-13-two-stage (2019-2023) CARDIATEAM project on Diabetic Cardiomyopathy (P.I. di Unità)

- Riconoscimenti e Premi**
- Chairman e organizzatore della sessione "Malattie Complesse" al congresso della Società Italiana di Genetica Umana (SIGU) 2009, Torino, Italia.
 - Chairman e organizzatore della sessione "Genomica Funzionale" al congresso della Società Italiana di Genetica Umana (SIGU) 2013, Milano, Italia.
 - Chairman e organizzatore della Tavola Rotonda sul Network dei Genomi Italiani, al congresso della Società Italiana di Genetica Umana (SIGU) 2015, Rimini, Italia
 - Membro del Comitato Scientifico e Chairman della Società Italiana di Genetica Umana (SIGU) 2016, e organizzatore e chairman della Tavola Rotonda sul Network dei Genomi Italiani, Torino, Italia
 - Membro del Comitato Scientifico e relatore di diversi corsi nell'ambito della Scuola Medica Ospedaliera 2016-2017, s017-2018, 2018-2019 (21 ECM ciascuno); Geni e test genetici: dal laboratorio alle applicazioni cliniche. Istituto Mendel, Roma, Italia
- Membro di organizzazioni scientifiche**
- Membro della Società Italiana di Genetica Umana (SIGU)
 - Membro della European Society of Human Genetics (ESHG)
 - Membro della American Society of Human Genetics (ASHG)
 - Membro della American Association of Cancer Research (AACR)
 - Membro Comitato Esecutivo della Società Italiana di Biometria (SIB) (2000-2003)
 - Membro dello "Study Group on DNA repair" coordinato dal Prof. Kraemer K. (<http://sigs.nih.gov/DNA-repair/Pages/default.aspx>)
 - Membro dell'International Consortium on Bladder Cancer (ICBC)
 - Membro dell'International Mesothelioma Interest Group (IMIG)
 - Membro del Centro Interdipartimentale "G. Scansetti" per lo studio dell'asbesto e di altre fibre tossiche.
 - Membro e Fondatore del Network for Italian Genomes (NIG; <http://www.nig.cineca.it/>)

ULTERIORI INFORMAZIONI

Metrica pubblicazioni IF TOTALE: 1137.19
IF MEDIO: 6.39
H-INDEX: 46

- Libri**
1. Torres SM, Erdei E, Berwick M, **MATULLO G**, Vineis P. (2012) Molecular Epidemiology. Chap. 4. In Handbook of Epidemiology, 2nd Edition, Springer Publishers.
 2. Paolo Vineis, GIUSEPPE MATULLO, Marianne Berwick. (2004) Molecular Epidemiology. Chap. 7. In Handbook of Epidemiology, John M. Last, Springer Publishers.
 3. Berwick M, **MATULLO G**, Vineis P. (2002) Studies of DNA repair and human cancer: an update. Chap.7: 83-107. In Biomarkers of environmentally associated disease: technologies, concepts, and perspectives. Edited by Wilson SH and Suk WA. Lewis publishers. CRC Press LLC, New York.
 4. Colajanni E, **MATULLO G**, Piazza A. (1997) Genetica e malattie cardiovascolari. In Selezione di Argomenti di Cardiologia: per i candidati all'idoneità apicale. Bergerone S e Morello M. Centro Scientifico Editore. Volume 2; pp. 1-12.
 5. English translation for ADELPHI Editor, chapters 1 e 2 of the book: "The History and Geography of Human Genes". Cavalli-Sforza LL, Menozzi P, Piazza A. Princeton, Princeton University Press, 1994.

- Pubblicazioni**
1. Li C, Stoma S, Lotta LA, Warner S, Albrecht E, Allione A, Arp PP, Broer L, Buxton JL, Da Silva Couto Alves A, Deelen J, Fedko IO, Gordon SD, Jiang T, Karlsson R, Kerrison N, Loe TK, Mangino M, Milaneschi Y, Miraglio B, Pervjakova N, Russo A, Surakka I, van der Spek A, Verhoeven JE, Amin N, Beekman M, Blakemore AI, Canzian F, Hamby SE, Hottenga JJ, Jones PD, Jousilahti P, Mägi R, Medland SE, Montgomery GW, Nyholt DR, Perola M, Pietiläinen KH, Salomaa V, Sillanpää E, Suchiman HE, van Heemst D, Willemsen G, Agudo A, Boeing H, Boomsma DI, Chirlaque MD, Fagherazzi G, Ferrari P, Franks P, Gieger C, Eriksson JG, Gunter M, Hägg S, Hovatta I, Imaz L, Kaprio J, Kaaks R, Key T, Krogh V, Martin NG, Melander O, Metspalu A, Moreno C, Onland-Moret NC, Nilsson P, Ong KK, Overvad K, Palli D, Panico S, Pedersen NL, Penninx BWJH, Quirós JR, Jarvelin MR, Rodríguez-Barranco M, Scott RA, Severi G, Slagboom PE, Spector TD, Tjonneland A, Trichopoulou A, Tumino R, Uitterlinden AG, van der Schouw YT, van Duijn CM, Weiderpass E, Denchi EL, **MATULLO G**, Butterworth AS, Danesh J, Samani NJ, Wareham NJ, Nelson CP, Langenberg C, Codd V. Genome-Wide Association Analysis in Humans Links Nucleotide Metabolism to Leukocyte Telomere Length. Am J Hum Genet. 2020 Feb 20. pii: S0002-9297(20)30048-3. doi: 10.1016/j.ajhg.2020.02.006. [Epub ahead of print] PubMed PMID: 32109421.
 2. Cocca M, Barbieri C, Concas MP, Robino A, Brumat M, Gandin I, Trudu M, Sala CF, Vuckovic D, Giroto G, **MATULLO G**, Polasek O, Kolčič I, Gasparini P, Soranzo N, Toniolo D, Mezzavilla M. A bird's-eye view of Italian genomic variation through whole-genome sequencing. Eur J Hum Genet. 2019

Nov 29. doi: 10.1038/s41431-019-0551-x. [Epub ahead of print] PubMed PMID: 31784700.

3. Antonio ML, Gao Z, Moos HM, Lucci M, Candilio F, Sawyer S, Oberreiter V, Calderon D, Devitofranceschi K, Aikens RC, Aneli S, Bartoli F, Bedini A, Cheronet O, Cotter DJ, Fernandes DM, Gasperetti G, Grifoni R, Guidi A, La Pastina F, Loreti E, Manacorda D, **MATULLO G**, Morretta S, Nava A, Fiocchi Nicolai V, Nomi F, Pavolini C, Pentiricci M, Pergola P, Piranomonte M, Schmidt R, Spinola G, Sperduti A, Rubini M, Bondioli L, Coppa A, Pinhasi R, Pritchard JK. Ancient Rome: A genetic crossroads of Europe and the Mediterranean. *Science*. 2019 Nov 8;366(6466):708-714. doi: 10.1126/science.aay6826. PubMed PMID: 31699931.

4. 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prof. Giuseppe Matullo

Dati personali

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