



CURRICULUM VITAE

PROF. GIOVANNI PORTA



INFORMAZIONI PERSONALI

Nome	GIOVANNI PORTA
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Nazionalità	Italiana
Data di nascita	[20-04-1962]

ESPERIENZA LAVORATIVA

- 2016 -2026 Membro Direzione Scientifica ASM (Associazione Italiana Studio Malformazioni)
- 2019 Segretario Direzione Scientifica ASM (Associazione Italiana Studio Malformazioni)
- 2018-2026 Consulente Clinico e di ricerca in Genetica Medica Centro Diagnostico Italiano
- 2017-2026 Consulente Specialista in Genetica Medica della rivista "Bimbi sani e belli".
- 2017-2026 Responsabile del Filo Rosso della Genetica collaborazione tra Uninsubria e ASM per la consulenza genetica telefonica, unico punto italiano.
- 2019-2026 Delegato all'Internazionalizzazione Corso di Laurea di Medicina e Chirurgia.
- 2006-2026 Professore Associato in Genetica Medica Università dell'Insubria, Varese
- 2010- 2026 Direttore della scuola di Specialità in Genetica Medica Uninsubria, Varese.
- 2012 Visiting Professor a tempo determinato MRC Leicester Toxicology Unit , UK
- 1999-2001 Visiting Professor , NIA, NIH, Baltimore, MD, USA
- 1992-2006 Ricercatore II Facoltà di Medicina e Chirurgia Università di Pavia, sede Varese
- 1993-1999 Consulente di Genetica Medica presso la Clinica di Ostetricia e Ginecologia Ospedale San Paolo
- 1988-1994 Post Dottorato Center for Genetic in Medicine Washington University, St Louis, Mo USA (Human Genome Project).

ISTRUZIONE E FORMAZIONE

1982-1988 Laurea in Medicina e Chirurgia Università di Pavia Tesi “ Effetto di dosaggio genico in cellule con monosomia del cromosoma 22 derivate da meningiomi umani” Relatore Marco Fraccaro con massimo dei voti.

1992 Specialità in Genetica Medica Dipartimento di Patologia Umana ed Ereditaria Università di Pavia, Tesi “ Fingerprinting basato sull'uso di sequenze ripetute per l'ordinamento di YACs in Xq2.4-Xq2.8.” Massimo dei voti e lode

Inglese

Eccellente

Progetti in corso

La ricerca del Prof Porta si focalizza sulle basi molecolari del differenziamento cellulare nei processi neoplastici , pre-neoplastici, infiammatori e fisiologici con particolare interesse al pathway p53-OTX.

La patogenesi della Leucemia Mieloide Cronica è un argomento di studio per la valutazione di protocolli di interruzione della terapia attraverso il monitoraggio della malattia minima residua con bio-marcatore basati sul DNA.

La valutazione del batterioma umano attraverso tecniche di sequenziamento di nuova generazione.

Valutazione del DNA circolante nelle patologie neoplastiche e setticemiche.

Analisi molecolari delle mielo-displasia somatiche ed acquisite.

Studio di farmaci che inducono il read-through traduzionale come nuove opzioni terapeutiche per i pazienti con anemia di Fanconi.

Docenze

Docente del corso di Scienze della vita e in particolare di Genetica e Biologia del corso di laurea in scienze infermieristiche dal 1994 al 2026

Docente del corso di Scienze della vita e in particolare di Genetica e Biologia del corso di laurea in fisioterapia dal 1994 al 2026.

Docente del corso di Scienze della vita e in particolare di Genetica e Biologia del corso di laurea in scienze motorie dal 1994 al 2026.

Docente del corso di Scienze della vita e in particolare di Genetica e Biologia del corso di laurea in odontoiatria dal 2015 al 2026.

Docente del corso di Genetica Medica e Biologia (I anno) del corso di laurea in Medicina e Chirurgia dal 2017 al 2025.

Docente del corso di Genetica Medica (VI anno) del corso di laurea in Medicina e Chirurgia dal 2017 al 2026.

Docente in vari Corsi di Scuola di Specialita' in area Medica 1994-2026.

Pubblicazioni:

Development of translational read-through-inducing drugs as novel therapeutic options for patients with Fanconi anemia

Hristodor, A.M., Cappelli, E., Baldisseri, E., ... Cipolli, M., Bezzerri, V.
Cell Death DiscoveryOpen source preview, 2025, 11(1), 286

Ataluren improves hematopoietic and pancreatic disorders in Shwachman-Diamond syndrome patients: a compassionate program case-series

Bezzi, V., Pegoraro, A., Hristodor, A.M., ... Corey, S.J., Cipolli, M.
Nature CommunicationsOpen source preview, 2025, 16(1), 8189

A Distinctive Type of Mosaic Variegated Aneuploidy: Case Report and Review of the Literature

Frattoni, A., Micheloni, G., Musio, A., ... Pasquali, F., Valli, R.
American Journal of Medical Genetics Part AOpen source preview, 2025, 197(2), e63901

Shwachman-Diamond Syndrome and Diabetes: An Update from the Italian Registry and Review of the Literature. Minelli, A., Pintani, E., Valli, R., ... Cipolli, M., Danesino, C.

Experimental and Clinical Endocrinology and DiabetesOpen source preview, 2025, 133(2), pp. 78–8

The synergism of SMC1A cohesin gene silencing and bevacizumab against colorectal cancer

Di Nardo, M., Astigiano, S., Baldari, S., ...Soddu, S., Musio, A.

Behavioral Alterations of Spatial Cognition and Role of the Apolipoprotein E-ε4 in Patients with MCI Due

to Alzheimer's Disease: Results from the BDSC-MCI Project

Cammisuli, D.M., Bellocchio, V., Milesi, A., ... Mauro, A., Castelnuevo, G.

Journal of Clinical MedicineOpen source preview, 2024, 13(18), 5447

Ten Years of Experience With a Telemedicine Platform Dedicated to Health Care Personnel:

Implementation Report

Azzolini, C., Premi, E., Donati, S., ...Giardina, G., Paolucci, U.

JMIR Medical Informatics, 2024, 12(1), e42847

Ataluren improves myelopoiesis and neutrophil chemotaxis by restoring ribosome biogenesis and reducing p53 levels in Shwachman–Diamond syndrome cells

Cipolli, M., Boni, C., Penzo, M., ...Polini, A., Bezzerri, V.

British Journal of Haematology, 2024, 204(1), pp. 292–305

OTX Genes in Adult Tissues

Terrinoni, A., Micheloni, G., Moretti, V., ...Valli, R., Porta, G.

International Journal of Molecular Sciences, 2023, 24(23), 16962

Donor Cell Acute Myeloid Leukemia after Hematopoietic Stem Cell Transplantation for Chronic Granulomatous Disease: A Case Report and Literature Review

Micheloni, G., Frattini, A., Donini, M., ...Valli, R., Pasquali, F.

Genes, 2023, 14(11), 2085

The Potential Role of the T2 Ribonucleases in TME-Based Cancer Therapy

Campomenosi, P., Mortara, L., Bassani, B., ...Bruno, A., Acquati, F.

Biomedicines, 2023, 11(8), 2160

Occurrence of L1M Elements in Chromosomal Rearrangements Associated to Chronic Myeloid Leukemia (CML): Insights from Patient-Specific Breakpoints Characterization

L'Abbate, A., Moretti, V., Pungolino, E., ...Porta, G., Cairoli, R.

Genes, 2023, 14(7), 1351

Apoptotic cell death in disease- Current understanding of the NCCD 2023

Vitale, I., Pietrocola, F., Guilbaud, E., ...Kromer, G., Galluzzi, I.

Cell Death and Differentiation, 2023,30(5),pp.1097-1154.

Human RNASET2: A Highly Pleiotropic and Evolutionary Conserved Tumor Suppressor Gene Involved in the Control of Ovarian Cancer Pathogenesis.

Bruno A, Noonan DM, Valli R, Porta G, Taramelli R, Mortara L, Acquati F. Int J Mol Sci. 2022 Aug

13;23(16):9074. doi: 10.3390/ijms23169074. PMID: 36012339

Case Report: Heterozygous Germline Variant in EIF6 Additional to Biallelic SBDS Pathogenic Variants in a Patient With Ribosomopathy Shwachman-Diamond Syndrome.

Taha I, Foroni S, Valli R, Frattini A, Rocca P, Porta G, Zecca M, Bergami E, Cipolli M, Pasquali F, Danesino C, Scotti C, Minelli A. Front Genet. 2022 Aug 12;13:896749. doi: 10.3389/fgene.2022.896749.

eCollection 2022. PMID: 36035165

Soybean Meal-Dependent Acute Intestinal Inflammation Delays Osteogenesis in Zebrafish Larvae.

Carnovali M, Banfi G, Porta G, Mariotti M. Int J Mol Sci. 2022 Jul 5;23(13):7480. doi:

10.3390/ijms23137480. PMID: 35806483

Macular Holes: Main Clinical Presentations, Diagnosis, and Therapies.

Premi E, Donati S, Azzi L, Porta G, Metrangolo C, Fontanel L, Morescalchi F, Azzolini C. J Ophthalmol.

2022 Apr 11;2022:2270861. doi: 10.1155/2022/2270861. eCollection 2022. PMID: 35450323

Soy diet induces intestinal inflammation in adult Zebrafish: Role of OTX and P53 family.

Micheloni G, Carnovali M, Millefanti G, Rizzetto M, Moretti V, Montalbano G, Acquati F, Giaroni C, Valli R, Costantino L, Ferrara F, Banfi G, Mariotti M, Porta G. Int J Exp Pathol. 2022 Feb;103(1):13-22. doi:

10.1111/iep.12420. Epub 2021 Nov 1. PMID: 34725870

Expression of Otx Genes in Müller Cells Using an In Vitro Experimental Model of Retinal Hypoxia. Azzolini C, Donati S, Micheloni G, Moretti V, Valli R, Acquati F, Costantino L, Ferrara F, Borroni D, Premi E, Testa F, Simonelli F, Porta G. *J Ophthalmol.* 2021 Dec 31;2021:6265553. doi: 10.1155/2021/6265553. eCollection 2021. PMID: 35003791

Enhanced p53 Levels Are Involved in the Reduced Mineralization Capacity of Osteoblasts Derived from Shwachman-Diamond Syndrome Subjects.

Frattini A, Bolamperti S, Valli R, Cipolli M, Pinto RM, Bergami E, Frau MR, Cesaro S, Signo M, Bezzeri V, Porta G, Khan AW, Rubinacci A, Villa I. *Int J Mol Sci.* 2021 Dec 11;22(24):13331. doi: 10.3390/ijms222413331. PMID: 34948128

The frequent and clinically benign anomalies of chromosomes 7 and 20 in Shwachman-diamond syndrome may be subject to further clonal variations.

Khan AW, Kennedy A, Furutani E, Myers K, Frattini A, Acquati F, Roccia P, Micheloni G, Minelli A, Porta G, Cipolli M, Cesaro S, Danesino C, Pasquali F, Shimamura A, Valli R. *Mol Cytogenet.* 2021 Nov 24;14(1):54. doi: 10.1186/s13039-021-00575-w. PMID: 34819134

Cohesin Mutations Induce Chromatin Conformation Perturbation of the H19/IGF2 Imprinted Region and Gene Expression Dysregulation in Cornelia de Lange Syndrome Cell Lines.

Pileggi S, La Vecchia M, Colombo EA, Fontana L, Colapietro P, Rovina D, Morotti A, Tabano S, Porta G, Alcalay M, Gervasini C, Miozzo M, Sirchia SM. *Biomolecules.* 2021 Nov 2;11(11):1622. doi: 10.3390/biom11111622. PMID: 34827619

Soybean Meal-Dependent Intestinal Inflammation Induces Different Patterns of Bone-Loss in Adult Zebrafish Scale.

Carnovali M, Valli R, Banfi G, Porta G, Mariotti M. *Biomedicines.* 2021 Apr 6;9(4):393. doi: 10.3390/biomedicines9040393. PMID: 33917641

Identification of OTX1 and OTX2 As Two Possible Molecular Markers for Sinonasal Carcinomas and Olfactory Neuroblastomas.

Micheloni G, Millefanti G, Conti A, Pirrone C, Marando A, Rainero A, Tararà L, Pistochini A, Lo Curto F, Pasquali F, Castelnuovo P, Acquati F, Grimaldi A, Valli R, Porta G. *J Vis Exp.* 2019 Feb 28;(144). doi: 10.3791/56880. PMID: 30882801

A Cell-Autonomous Oncosuppressive Role of Human RNASET2 Affecting ECM-Mediated Oncogenic Signaling. Roggiani F, Riva C, Raspagliesi F, Porta G, Valli R, Taramelli R, Acquati F, Mezzanzanica D, Tomassetti A. *Cancers (Basel).* 2019 Feb 22;11(2). pii: E255. doi: 10.3390/cancers11020255. PMID: 30813308

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D'Amico G, Frattini A, Montalbano G, Khan AW, Porta G, Millefanti G, Olivieri C, Cipolli M, Cesaro S, Pasquali F, Danesino C, Cazzaniga G, Maserati E. *Br J Haematol.* 2019 Mar;184(6):974-981. doi: 10.1111/bjh.15729. Epub 2018 Dec 26. PMID: 30585299

gDNA qPCR is statistically more reliable than mRNA analysis in detecting leukemic cells to monitor

CML. Rainero A, Angaroni F, D'Avila F, Conti A, Pirrone C, Micheloni G, Tararà L, Millefanti G, Maserati E, Valli R, Spinelli O, Buklijas K, Michelato A, Casalone R, Barlassina C, Barcella M, Sirchia S, Piscitelli E, Caccia M, Porta G. *Cell Death Dis.* 2018 Mar 2;9(3):349. doi: 10.1038/s41419-018-0387-2.

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neuroblastomas. Pirrone C, Chiaravalli AM, Marando A, Conti A, Rainero A, Pistochini A, Lo Curto F, Pasquali F, Castelnuovo P, Capella C, Porta G. *Eur J Histochem.* 2017 Feb 9;61(1):2730. doi: 10.4081/ejh.2017.2730. PMID: 28348423

Nitric oxide regulates homeoprotein OTX1 and OTX2 expression in the rat myenteric plexus after

intestinal ischemia-reperfusion injury. Filpa V, Carpanese E, Marchet S, Pirrone C, Conti A, Rainero A, Moro E, Chiaravalli AM, Zucchi I, Moriondo A, Negrini D, Crema F, Frigo G, Giaroni C, Porta G. *Am J Physiol Gastrointest Liver Physiol.* 2017 Apr 1;312(4):G374-G389. doi: 10.1152/ajpgi.00386.2016. Epub 2017 Feb 2

Macular hole surgery: the healing process of outer retinal layers to visual acuity recovery. Caprani SM, Donati S, Bartalena L, Vinciguerra R, Mariotti C, Testa F, Porta G, Azzolini C. *Eur J Ophthalmol*. 2017 Mar 10;27(2):235-239. doi: 10.5301/ejo.5000905. Epub 2017 Feb 22. PMID: 28233890

OTX2 regulates the expression of TAp63 leading to macular and cochlear neuroepithelium development. Porta G Palombo R, Bruno E, Provero P, Serra V, Neduri K, Viziano A, Alessandrini M, Micarelli A, Ottaviani F, Melino G, Terrinoni A. *Aging (Albany NY)*. 2015 Nov;7(11):928-36. PMID: 26554466

gDNA Q-PCR for clinical monitoring of CML Porta G, Pagani IS, Pirrone C. *Cell Cycle*. 2015;14(23):3659-60. doi: 10.1080/15384101.2015.1006053.

Genomic quantitative real-time PCR proves residual disease positivity in more than 30% samples with negative mRNA-based qRT-PCR in Chronic Myeloid Leukemia. Ilaria S. Pagani, Orietta Spinelli, Elia Mattarucchi, Cristina Pirrone, Diana Pigni, Elisabetta Amelotti, Silvia Lilliu, Chiara Boroni, Tamara Intermesoli, Ursula Giussani, Luigi Caimi, Federica Bolda, Renata Baffelli, Eleonora Candi, Francesco Pasquali, Francesco Lo Curto, Arnalda Lanfranchi, Fulvio Porta, Alessandro Rambaldi and Giovanni Porta, *Oncoscience*, vol1, no7, 510-521

Vitreous substitutes: the present and the future. Donati S, Caprani SM, Airaghi G, Vinciguerra R, Bartalena L, Testa F, Mariotti C, Porta G, Simonelli F, Azzolini C. *Biomed Res Int*. 2014;2014:351804. doi: 10.1155/2014/351804. Epub 2014 May 4

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Metabolic effects of TiO2 nanoparticles, a common component of sunscreens and cosmetics, on human keratinocytes. Tucci P, Porta G, Agostini M, Dinsdale D, Iavicoli I, Cain K, Finazzi-Agró A, Melino G, Willis A. *Cell Death Dis*. 2013 Mar 21;4:e549. doi: 10.1038/cddis.2013.76. PMID: 23519118 IF 6.044

Expression of VEGF-A, Otx Homeobox and p53 Family Genes in Proliferative Vitreoretinopathy Claudio Azzolini,¹ Ilaria Stefania Pagani,² Cristina Pirrone,^{2,3} Davide Borroni,^{1,2} Simone Donati,¹ Muna Al Oum,¹ Diana Pigni,² Anna Maria Chiaravalli,⁴ Riccardo Vinciguerra,¹ Francesca Simonelli,⁵ and Giovanni Porta² *Mediators of Inflammation* Volume 2013 (2013), Article ID 857380 IF 3.882

Differential signature of the centrosomal MARK4 isoforms in glioma. Magnani I, Novielli C, Fontana L, Tabano S, Rovina D, Moroni RF, Bauer D, Mazzoleni S, Colombo EA, Tedeschi G, Monti L, Porta G, Bosari S, Frassoni C, Galli R, Bello L, Larizza L. *Anal Cell Pathol (Amst)*. 2011;34(6):319-38. doi: 10.3233/ACP-2011-0031. PMID: 22156016 IF 0.9

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The mammary gland and the homeobox gene Otx1. Pagani IS, Terrinoni A, Marengi L, Zucchi I, Chiaravalli AM, Serra V, Rovera F, Sirchia S, Dionigi G, Miozzo M, Frattini A, Ferrari A, Capella C, Pasquali F, Lo Curto F, Albertini A, Melino G, Porta G. *Breast J*. 2010 Sep-Oct;16 Suppl 1:S53-6. doi: 10.1111/j.1524-4741.2010.01006.x. IF 1.831

Molecular monitoring of residual disease in chronic myeloid leukemia by genomic DNA compared with conventional mRNA analysis. Mattarucchi E, Spinelli O, Rambaldi A, Pasquali F, Lo Curto F, Campiotti L, Porta G. *J Mol Diagn*. 2009 Sep;11(5):482-7. doi: 10.2353/jmoldx.2009.080150. PMID: 19710400 IF 3.770

Misbehaviour of XIST RNA in breast cancer cells. Sirchia SM, Tabano S, Monti L, Recalcati MP, Gariboldi M, Grati FR, Porta G, Finelli P, Radice P, Miozzo M.

PLoS One. 2009;4(5):e5559. doi: 10.1371/journal.pone.0005559. Epub 2009 May 15.
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Mattarucchi E, Guerini V, Rambaldi A, Campiotti L, Venco A, Pasquali F, Lo Curto F, Porta G.
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Porta G, Maserati E, Mattarucchi E, Minelli A, Pressato B, Valli R, Zecca M, Bernardo ME, Lo Curto F,
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Porta G, Mattarucchi E, Maserati E, Pressato B, Valli R, Morerio C, Zecca M, Panarello C, Locatelli F, Lo
Curto F, Pasquali F.
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Establishment and study of different real-time polymerase chain reaction assays for the quantification of
cells with deletions of chromosome 7.
Mattarucchi E, Marsoni M, Passi A, Lo Curto F, Pasquali F, Porta G.
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Two patients with COMT inhibitor-induced hepatic dysfunction and UGT1A9 genetic polymorphism.
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development, characterization and pre-validation.
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breast cancer cells.
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Karousou EG, Porta G, De Luca G, Passi A.
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HIV-1 proviral DNA polymerase chain reaction detection in chorionic villi after exclusion of maternal contamination by variable number of tandem repeats analysis. De Anderis C, Simoni G, Rossella F, Castagna C, Pesenti E, Porta G, Colucci G, Giustelli S, Pardi G, Semprini AE. *AIDS* (1996) 10(7):711-5. PMID: 8805861 UI: 9639935

Human CD40L gene maps between DXS144E and DXS300 in Xq26. Pilia G, Porta G, Padayachee M, Malcolm S, Zucchi I, Villa A, Macchi P, Vezzoni P, Schlessinger D. *Genomics* (1994) 22:249-251. PMID: 7959785 UI: 95048387 I.F. 4,0

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Prof. Giovanni Porta

A handwritten signature in black ink, appearing to be 'G. Porta' or similar, written in a cursive style.