PERSONAL INFORMATION

Paolo Gasparini, CF: GSPPLA60R25G224L

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Nationality Italian

Accomplished Consultant, Researcher and Professor of Medical Genetics at the University of Trieste, Head of Medical Genetics Service and Head of the Department for Advanced Diagnostics and Clinical Trials at the Institute for Maternal and Child Health IRCCS Burlo Garofolo in Trieste.
Amongst other prestigious assignments, he is the President of the Italian Society of Human Genetics (SIGU), Member (Representative of the European Clinicians) of the Committe for Advanced Therapies (CAT) of the European Medicine Agency (EMA) and Italian Delegate of the EMA's Committee for Medicinal Products for Human Use (CHMP).

WORK EXPERIENCE

11/2010–Present — Head of the Department of Advanced Diagnostics and Clinical Research at the Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste

IRCCS materno infantile Burlo Garofolo Trieste, Trieste (Italy)

06/2005–Present — Full Professor of Medical Genetics at the University of Trieste

University of Trieste, Trieste (Italy)

Full professor of Medical Genetics.

Deputy Head of Department, Department of Medicine, Surgery and Health Sciences.

Director of the Medical Genetics Residency Program.

Coordinator of the Graduate School in Reproduction and Developmental Sciences.

From the beginning of 2015 until February 2017: Head of the Experimental Genetics Division, Research Department, Sidra Medical and Research Center, Doha, Qatar (during sabbatical leave).

06/2005–Present — Head of the Medical Genetics Service

IRCCS materno infantile Burlo Garofolo, Trieste (Italy)

Consultant in Genetics at the Genetics Department of the Institute for Maternal and Child Health IRCCS "Burlo Garofolo"in Trieste.

From 1 June 2009 to 30 June 2010 – Ad interim Scientific Director of the Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste.

He carries out an intense activity both within the hospital and also for external structures:

- He has contributed to the identification of the following disease genes: mitochondrial disease due to nuclear gene (2006), deafness due to PMC2A/Cadherin (2007), and Severe Infantile Encephalomyopathy (2008).
- He is responsible for the Burlo-CBM (Consorzio per il Centro di Biomedicina Molecolare) genomic facilities (recognized by Telethon charity as the national referral center for high-throughput SNPs genotyping).
- He is one of the founders of the Italian Network on Genetic Isolates (INGI) and is directly involved in two large studies on isolated populations (Carlantino Project and FVG Genetic Park Project) in which more than 4,000 individuals were identified, accurately phenotyped and genotyped.
- He is involved in several national and international collaborations on technological aspects of DNA analysis (including lab-on-a chip development), biobanking, and analysis of complex and quantitative traits including LATEMAR, PHOEBE, and TECHGENE consortia

from EU.

- In 2009 he was appointed as Visiting Scientist at the Sanger Center (Cambridge, UK) in collaboration with the group lead by Prof. L. Peltonen. Thanks to his work, INGI network entered the International Consortia on complex and quantitative traits, and became one of the Italian prototypes on biobanking to BBMRI.
- In 2010 he planned and organised a phenotype and sample collection of populations across the Silk Road (www.marcopolo2010.it).

His most recent research interests include the genetic foundations of hereditary and complex diseases, with a specific focus on sense organs and related diseases (e.g. hearing, taste, food preferences, smell).

06/2005–Present **Prestigious assignments and offices**

- Member of the AIFA Research & Development Working Group.
- Member of the National Committee of Biotechnology, Biosafety and Life Sciences of the Italian Government (CNBBSV).
- Chairman of the Scientific Advisory Committee of IDIBELL de l'Hospitalet de Llobregat (Barcelona, Spain), of which he was previously a member.
- From January 2012 to March 2021 he was a member (Italian Delegate) of the Committee of the Agency for Advanced Therapies (CAT) of the European Medical Agency (EMA).
- In January 2020 he was appointed Expert of the Working Group "Transfer of Genomic Therapy into Clinical Practice" (Esperto del Gruppo di lavoro "Trasferimento della Terapia Genomica nella pratica clinica") by the President of the Superior Council of Health.
- In 2022 he was appointed Member of the National Committee for Rare Diseases, member of the National Coordination Group for Bioeconomy, member of the Technical Table on Advanced Therapies established by the Ministry of Health.
- In July 2022 he was appointed Member (Representative of European Doctors) of the Committee for Advanced Therapies (CAT) EMA.
- In November 2022 he was elected President of the Italian Society of Human Genetics (SIGU), of which he was previously a member of the Board of Directors.
- In 2023 he was appointed Italian delegate of the CHMP (Committee for Medicinal Products for Human Use) of EMA.
- In September 2023 he was appointed member of the Scientific Committee of OMAR: Osservatorio Malattie Rare.

11/2001–06/2005 **Associate professor of Medical Genetics at the Faculty of Medicine at the SecondUniversity of Naples**

University of Naples, Naples (Italy)

He founded the Medical Genetics Laboratory within the General Pathology Department.

He registered within the Centre of Excellence for Cardiovascular Diseases of the Second University of Naples. There, he started a Nanotechnology laboratory. He was also in charge of the Linkage and Mapping Unit of the TIGEM Institute (Telethon Institute of Genetics and Medicine) in Naples and coordinated a research group within TIGEM. He carried on his research activities on hearing loss and on the definition of molecular bases of multi-factor diseases in genetically isolated populations. During this time, he contributed to the identification of genes for the following diseases: hearing loss caused by myosin 1, hearing loss caused by myosin MYH14, hearing loss caused by Espin gene mutations, methylmalonic encephalopathy.

He was Head of the Medical Genetics Doctoral School from October 2003 to June 2005.

In November 2003 he was appointed Head of the Medical Genetics Service at the Laboratory Medicine Department of the Second University of Naples University Hospital. As Head of Service, he established and managed the Genetic Consulting Service, the Molecular Diagnostic Laboratory and all cytogenetic activities, introducing new services that were urgently needed.

He was the representative of the University Hospital within the regional network of assistance in favour of patients affected by rare diseases. He started SUN-GENS, the first national SNP genotyping service, which worked with many Italian Universities.

03/1993–10/2001 **Senior Physician at the Medical Genetics Service**

Ospedale Casa Sollievo della Sofferenza, San Giovanni Rotondo (Italy)

He carried on research and diagnostics on hereditary hemochromatosis, on Type 1 neurinomatosis, on cystic fibrosis, on Duchenne's dystrophy and on the adult policystic kidney.

He worked on a project on cystinic calculosis with a decisive contribution to the detection of the gene that causes such disease once it is altered (April 1994). He contributed to demonstrating the presence of genetic heterogeneity. He also contributed to the detection of a gene causing rare syndromes, including cataract and hyperferritinemia (1995). He conducted an important research project on genetic deafness and identified connexin 26 as the gene causing the most common type of genetic deafness (1997).

In 1998 he collaborated on a research project leading to the identification of the gene of Leigh's disease.

From July to October 1998 he was Visiting Professor at the Department of Haematology at the University of Pennsylvania, Philadelphia (USA). There he acquired the basic knowledge for the development of the new microchip technologies. I

In 1999 he coordinated researchers of the Medical Genetics Department who cloned and identified connexin 30, a new gene for deafness. He also significantly contributed to the identification of the gene of non-I cystic renal calculosis.

In 2000 he contributed to the identification of a new gene causing hereditary hemochromatosis (TFR2).

In 2001 he contributed to the identification of the following genes: BPES syndrome, deafness caused by gene myosin 6, type III Usher Syndrome, dominant hemochromatosis (HFE4).

In this period, he was also involved in the management of the Mapping Group of the Medical Genetics Service, thus leading to the identification of several locus diseases.

The results of this intense research activity were shared in many papers presented at national and international meetings and congresses.

He started several research projects on the molecular bases of multi-factor diseases. In particular, he was involved in research projects on celiac disease, Chron's disease and ulcerative rectocolitis, osteoporosis. He also set up a project for the study of a geographic genetic isolates (Carlantino Project).

During this period, he was also Genetic Consultant (Medical and Clinical) at the Medical Genetics Service and he managed the Molecular Diagnostics section.

05/1992–03/1993 **Medical Assistant at the Medical Genetics Service**

Ospedale Casa Sollievo della Sofferenza, San Giovanni Rotondo (Italy)

Management of the Medical Genetics Laboratory. He managed the Laboratory and started several research projects on hereditary hemochromatosis, cystic fibrosis and neurinomatosis. He also contributed to the development and implementation of molecular biology techniques to the diagnosis of hereditary diseases such as Duchenne's muscular dystrophy and the adult polycystyc kidney. The results have been presented at several national and international conferences.

He was also Genetic Consultant (Medical and Clinical) at the Medical Genetics Service and managed the molecular diagnostics section by implementing the molecular diagnostics of several hereditary diseases.

02/1990–05/1992 **Researcher**

Institute of Biological Sciences of the University of Verona, Verona (Italy)

He actively participated to the discovery of several mutations withing the genes of cystic fibrosis, as well as to a deep analysis of the frequency of several other mutations of the gene within the Mediterranean Basin. These results were presented in a Lecture entitled "Cystic Fibrosis' screening in Italy: prospects after one year of the gene's discovery" held in Perugia in October 1990 within the session on the Screening of Genetic Diseases in Italy of the Joint National Meeting A.G.I. – F.I.S.M.E.

He researched the genetic defect that causes the development of a serious and common pathology such as primary hemachromatosis. A long, articulated analysis enabled the more precise detection of the gene causing such disease, within a small portion of chromosome 6.
The results of this research were presented at several national and international conferences.

02/1987–02/1990 Postdoctoral Fellow

Institute of Biological Sciences, Verona (Italy)

Researcher at the Institute of Biological Sciences in Verona.

a) Molecular analysis of the gene of Cystic Fibrosis, of the adult polycystyc kidney (APKD) and of the chronic obstructive bronchopneumopthy (BPCO)

b) Application of molecular probes to pre-natal diagnostics to detect CF carriers, development of rapid and sensitive techniques for the molecular analysis of several genetic diseases (DNA amplification, etc.)

d) Monitoring and follow-up of heterologous bone-marrow transplants

e) Analysis of the bcr-abl rearrangement in patients affected by Chronic Mieloid Leukaemia

f) Evaluation of the minimal residual disease (MRD)in patients affected by Chronic Mieloid Leucemia after interferon therapy and/or chemotherapy and/or bone marrow transplant

g) Characterization and development of molecular probes for individual identification purposes(forensic biology, etc.).
The results of the research were presented at several national and international conferences.

01/1985–01/1987 Postdoctoral Fellow

Department of Medical Sciences and Human Oncology of the University of Turin, Turin(Italy)

Attendance to the hospital ward and the laboratory. Main research in the study of molecular alterations in haematological diseases as well as in the study of oncogenes in the different pathologies, molecular genetics of thalassemias and leukemias.

Since 1986 he collaborated to the research programme "Polymorphism of human cellular oncogenes" at the National Research Centre in Turin, Italy.

PUBLICATIONS

He is author of more than 470 scientific publications.
ORCHID ID: https://orcid.org/0000-0002-0859-0856

SCOPUS ID: 22634397400

SCOPUS Index: 91

D Index: 94

EDUCATION AND TRAINING

1992 Residency in Medical Genetics

University of Verona (Italy)

Full marks cum laude.

1988 Residency in General Haematology (Clinical and Laboratory)

University of Verona (Italy)

Full marks cum laude.

1985 M.D. Degree

University of Turin (Italy)

Full marks cum laude.

1979 Secondary School Diploma in Classical Studies

Full marks .

ADDITIONAL INFORMATION

1. From February 2024: member of the Scientific Committee of the Graduate School of Health Economics and Management, one of the Graduate Schools of Università Cattolica.

2. From February 2024: member of the Scientific Committee of Lorenzini Foundation.

3. From September 2023: member of the National Committee for Rare Diseases (OMAR)

4. From 2023: Italian delegate of the EMA's Committee for Medicinal Products for Human Use (CHMP).

5. From November 2022: President of the Italian Society of Human Genetics (SIGU), where he was previously a member of the Board of Directors.

6. From July 2022: Member (Representative of the European Clinicians) of the Committe for Advanced Therapies (CAT) of the European Medicine Agency (EMA)

7. From January 2020: Expert of the Working Group "Transfer of Genomic Therapy to Clinical Practice" from the Consiglio Superiore di Sanità (CSS).

8. From June 2016: Member of the National Committee for Biosafety, Biotechnology and Life Sciences (CNBBSV).

9. From January 2012 to March 2021: Member (Italian Delegate) of the Committee of the Agency for Advanced Therapies (CAT) of the European Medical Agency (EMA).

10. From 2011 to 2013: Member of the AIFA Research & Development Working Group.

11. In January 2006 he was appointed member of the Technical-Scientific Commission of the CBM (Consortium for the Centre for Molecular Biomedicine) of Trieste, contributing to the start of the activities of the CBM itself. He collaborates with CBM in the direct management of the Genotyping Centre, as well as in the evaluation of the companies and activities of the CBM itself.

12. He is one of the founders of the "Italian Network on Genetic Isolates" (INGI).

13. He was a member of the Scientific Programme Committee of the European Society of Human Genetics (ESHG).

14. Chair of the IDIBELL Scientific Advisory Committee of the Hospitalet de Llobregat (Barcelona, Spain).

15. Advisor of the Board of Directors of the Italian Society of Human Genetics (Società Italiana di Genetica Umana - SIGU).

16. Organiser of several national and international scientific conferences, seminars and workshops.

Honours and awards

2021 Premio Medicina Italia for Scientific Research

2011 Premio Grande Ippocrate for Scientific Research

2010 Qatar National Research Foundation Award for Biomedicine

2003 The European Society of Human Genetics/Nature Publishing Group Award2000

International Journal of Molecular Medicine Award for Scientific Research 1999

Premio Augustale 99 Award for Scientific Research

1994 SIBIoC 94 Award

1991 AIRH Junior Award for Medical Genetics 1988

Socrea – Sirtori Award for Medical Genetics

1987-1990 The Cystic Fibrosis Foundation Fellowship

1986-1987 Gigi Ghiotti Foundation Fellowship

LANGUAGE SKILLS

Mother tongue(s) Italian

	Listening	Reading	Spoken interaction	Spoken production	Writing
English	C2	C2	C2	C2	C1
Spanish	C1	C1	C1	C1	B2