europass Curriculum vitae

PERSONAL INFORMATION

Paolo Gasparini

IRCCS Burlo Garofolo, Via dell'Istria, 65/1, 34137 Trieste (Italy)

+390403785540

paolo.gasparini@burlo.trieste.it

Nationality Italian

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.

Director of the Postgraduate School in Medical Genetics and 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 (on sabbatical time).

In January 2020 he has been appointed as 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 by the President of the Superior Council of Health.

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. During this period he has carried out an intense activity both within the hospital and also for external structures.

On 1 August 2007 he has been appointedHead of the Medical Genetics Laboratory of the Institute for Maternal and Child Health IRCCS "Burlo Garofolo", 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 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 was also responsible for the Burlo-CBM 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 4000 individuals were collected, accurately phenotyped and genotyped. He is further 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 spent some time as visiting scientist working at Sanger Center (Cambridge, UK) in collaboration with the group lead by Prof. L. Peltonen. Has brought INGI network within most of the competitive International Consortia on complex and quantitative traits, and successfully candidate it as one of the Italian prototypes on biobanking to BBMRI.

In 2010 he has planned and organised a phenotype and sample collection of populations across the Silk Road (www.marcopolo2010.it).

11/2001–06/2005

Associate professor in Medical Genetics at the Faculty of Medicine of the Second University of Naples

University of Naples, Naples (Italy)

He started the Medical Genetics Laboratory within the General Pathology Department. He also 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 also coordinated a research group within TIGEM. He carried on his research activities on hearing losses and on the definition of molecular bases of multi-factor diseases in genetically isolated populations. During this period 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.

Since November 2003 he was appointed Head of the Medical Genetics Service at the Laboratory Medicine Department of the Second University of Naples. University Hospital. Within this post he implemented and orgeanized the Genetic Consulting Service. The Molecular Diagnostic Laboratory and cytogenetic activities, thus introducing a structure that had been lacking up to November 2003.

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 continued research and diagnostics on hereditary hemochromatosis, on Type 1 neurinomatosis, on cystic fibrosis, on Duchenne's dystrophy and on the adult policystic kidney. He started a project on cystinic calculosis with a decisive contribution on the detection of the gene (April 1994), which causes such disease onece it is altered. He then contributed to demonstrate the presence of genetic heterogenity. He also contributed to the detection of a gene causing a rare syndrome including cataract and hyperferritinemia (1995). He started an important research project on genetic deafness and identified connexin 26 as the gene causing the most common type of genetic deafness (1997). During 1998 he collaborated at a research project leading to the identification of the gene of Leigh's disease and 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. In 1999 he coordinated researchers of the Medical Genetics Department who cloned and indentified connexin 30, a new gene for deafness.

In 1999 he gave a substantial contribution to the identification of the gene of non-l cystic renal calculosis. During 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 has also been 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 acivity have been the subject of several papers presented in many national and international meetings and congresses.

He also started some 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 started a project for the study of a geographic genetic isolates (Carlantino Project).

During this period he was also (Medical and Clinical) Genetic Consultant 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. During this period he organized the above mentioned lab 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 in several national and international

meetings

He was also (Medical and Clinical) Genetic Consultant 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 participated actively to the discovery of several mutations within the gene of cystic fibrosis

as well as to a deep analysis of the frequence of several other mutations of this

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 started to be involved in the research of the genetic defect involved in 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 have been presented in several national and international meetings.

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 have been presented in several national and international meetings

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.

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)

1979 **Secondary School Diploma in Classical Studies**
Full marks.

PERSONAL SKILLS

Mother tongue(s): Italian

Foreign language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	C2	C2	C2	C2	C1
Spanish	C1	C1	C1	C1	B2

Levels: A1 and A2: Basic user - B1 and B2: Independent user - C1 and C2: Proficient user
Common European Framework of Reference for Languages - Self-assessment grid

ADDITIONAL INFORMATION

In January 2006 he has been appointed as member of the technical-scientific committee of the Cluster in Biomedicine (CBM) and contributed to start the cluster's activities. He collaborated with CBM in the management of the Genotyping core facility as well as in the appraisal of companies and activities of the cluster.

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

Since January 2012: member of the Committee for Advanced Therapies (CAT) of the European Medicine Agency (EMA).

Member of the Scientific Advisory Board of IDIBELL of the Hospitalet de Llobregat (Barcelona, Spain).

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

Since June 2016: member of the National Committee of Biosecurity, Biotechnologies and Life Sciences (Comitato nazionale per la Biosicurezza, le Biotecnologie e le Scienze della Vita (Cnbbsv).

Honours and awards

2011 "Premio Grande Ippocrate"

2010 "Qatar National Research Foundation Award for Biomedicine"

2003 "The European Society of Human Genetics/Nature Publishing Group Award"

2000 "International Journal of Molecular Medicine Award" for scientific research

1999 "Premio Augustale 99" prize for scientific research

1994 "SIBIoC 94" award

1991 "AIRH Junior " prize for Medical Genetics

1988 "Socrea – Sirtori" prize for Medical Genetics

1987-1990 "The Cystic Fibrosis Fundation Fellowship"

1986-1987 "Gigi Ghiotti Fundation Fellowship"

Publications

He is author of more than 400 scientific publications.

ORCHID ID: https://orcid.org/0000-0002-0859-0856

SCOPUS ID: 22634397400