europass Curriculum vitae

PERSONAL INFORMATION Paolo Gasparini

WORK EXPERIENCE

January 2014–Present

Full Professor of Medical Genetics at the University of Trieste and Head of the Department of Advanced Diagnostics and Clinical Research at the Institute for Maternal and Child Health IRCCS “Burlo Garofolo”, Trieste

University of Trieste and IRCCS Burlo Garofolo Trieste (Italy)

From beginning 2015 until February 2017: Head of the Experimental Genetics Division, Research Department, Sidra Medical and Research Center, Doha, Qatar (on sabbatical time). From March 2017 back to University of Trieste and IRCCS-Burlo Garofolo.

June 2008–Present

Full Professor of Medical Genetics at the University of Trieste

University of Trieste (Italy)

Director of the Postgraduate School in Medical Genetics and Head of the Medical Genetics Laboratory at 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.

Since 1 November 2010 Head of the Department of Advanced Diagnostics and Clinical Research of the Institute for Maternal and Child Health IRCCS “Burlo Garofolo”, Trieste.

June 2005–May 2008

Full professor of Medical Genetics at the Faculty of Medicine of the University of Trieste, Department of Reproduction and Developmental Sciences

University of Trieste (Italy)

Professor of Medical Genetics at the Postgraduate School in Medical Genetics and also Director of the School since 2005.

Since 2005 he has also been a 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. 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. 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

November 2001–June 2005

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

University of 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.

March 1993–October 2001

Senior Physician at the Medical Genetics Service of the Scientific Institute "Ospedale Casa Sollievo della Sofferenza" in San Giovanni Rotondo (FG), Italy

Ospedale Casa Sollievo della Sofferenza (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. Gasparini also contributed to the detection of a gene causing a rare syndrome including cataract and hyperferritinemia (1995) More recently 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 15 July to 15 October he was visiting professor at the Department of Haematology of the University of Pennsylvania. 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-I 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.

May 1992–March 1993

Medical Assistant at the Medical Genetics Service of the Scientific Institute "Ospedale Casa Sollievo della Sofferenza" in San Giovanni Rotondo (FG), Italy

Ospedale Casa Sollievo della Sofferenza (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.

February 1990–May 1992

Researcher at the Institute of Biological Sciences of the University of Verona

University of 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

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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.

February 1987–February 1990

Researcher at the Institute of Biological Sciences in Verona

Institute of Biological Sciences (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

January 1984–January 1987

Resident doctor at the Clinical Medicine Deptartment of the University of Turin, Italy

University of 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. 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–

MD Degree

University of Turin (Italy)

1979–

Secondary School Diploma in Classical Studies

Full marks.

ADDITIONAL INFORMATION

Expertise

Publications

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Projects

Memberships

Other Relevant Information

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