CURRICULUM VITAE
STEFANO GAMBARDELLA

Rome, 29/10/1977
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EDUCATION

PhD Advanced Technology in Biomedicine, University of Rome Tor Vergata (2007-2011)
Specialist in Biochemical chemistry and molecular biology, University of Rome "Tor Vergata" (2002-2006)
Degree in Biological Sciences, University of Rome "Roma Tre" (1996-2002)

Personal Statement

I'm a molecular biologist with a strong expertise in Mendelian genetics, complex genetics, genomics, and functional genomics with a primary focus on Neurodegenerative and Cardiovascular diseases.

My research consider patient cohorts with a high genetic load and well-documented families with marked variability in disease penetrance and expression. In these patients I apply genetic and genomic approaches including whole-genome/exome sequencing (NGS), large-scale targeted re-sequencing, RNA sequencing to identify rare, highly penetrant mutations, risk and disease modifying genetic variations. Other studies investigate post-genomic consequences (e.g. transcriptomic, proteomic, metabolomics, epigenetic), environmental and lifestyle factors that also likely play a vital role in disease onset, penetrance and progression.

EMPLOYMENT

September 2012 – Today: Technical director of Molecular Genetic Centre, IRCCS Neuromed, Pozzilli, Italy
The service provides: i) diagnostic in Molecular Genetic, Molecular Cardiology, Pharmacogenetics, Medical Genetics, Infectious disease, Neurogenetics, Nutrigenetics, Molecular Oncology; II) Research on genetic component in neurological diseases (Alzheimer, Parkinson, Epilepsy, Multiple Sclerosis); III) Service in Sanger Sequencing and next generation sequencing

2010 – July 2012 Scientific consultant, Bios International, Research and Development Division, Rome, Italy
Patent drafting, *Research and application of public grant;* Scientific Project management SMEI-Universities; Industrial Project management, CE- Mark,

2009 – 2010: Researhcer for Bionoor, Pula, Italy
Assistant of production in Nutraceutical products (Nurax Corde, Nurax Redox, Nurax Flex, based on Nuragene patent).

2006 – 2009: Consultant for Technogenetics, Milano, Italy
Design of CGH array products (Gain or Loss Detection Kit, GOLD CHIP, version 1)

2001 – 2010: Fellow researcher, Tor Vergata University, Rome, Italy
Diagnosis of Genetic diseases (Prenatal and Postnatal diagnosis of Cystic Fibrosis; Postnatal diagnosis of Congenital deafness, Thomsen disease, Pancreatic disease, Tuberous sclerosis type 2)
Scientific Research in Genetic Area (Research activities in genetic, medical genetics and molecular biology; Research of modifier genes of Cystic Fibrosis; Research of new mutation in the genes responsible for Cystic Fibrosis Treacher Collins syndrome, X-linked anhidrotic ectodermal dysplasia, Acute recurrent pancreatitis, Hereditary angioedema; Gene expression analysis in Myotonic dystrophy, Cystic fibrosis, Mandibuloacral dysplasia; Implementation of genetic diagnosis on DHPLC; Study of miRNA in Myotonic dystrophy; Genetic analysis of phenotypic variability and segregation analysis; Set up of new array CGH protocol for diagnosis.
Tutor-Teaching activity

SKILLS

Experience in managing a Laboratory

Experience in managing a molecular genetics laboratory for research and diagnosis; experience in supervising students, technicians and assistants; management of personnel, materials, equipment, and funds in laboratories as well as business/industry setting

Experience in Project Management

General skills in management of scientific and informatics projects in research and industrial area; grant selection, fund raising, application, final reporting, coordination of the activities between public and private organizations

Experience in Diagnostic kit Design, Production and CE-IVD mark

Design, construction, validation, production of diagnostic kit in molecular biology and nutraceutical products, including patent draft and CE mark. Design of NGS panel for both diagnostic and research activity:

Design of CGH array products (Gain or Loss Detection Kit, GOLD CHIP, version 1, Technogenetics)

Design, production, validation and CE mark of DMD/BMD Detection kit for the diagnosis of Duchenne (DMD) adi Becker (BMD) dystrophies through multiplex PCR and capillary electrophoresis (Orga Bio Human srl)

Design, production, validation and CE mark of CX26- Cx30 Detection Kit for the diagnosis of most common mutation in genes coding for Connexins through multiplex PCR and capillary electrophoresis (Orga Bio Human srl)

Design, production, validation and CE mark of Thrombofil Detection Kit for the diagnosis of most common variation responsible of Thrombofilia through SNAPShot PCR and capillary electrophoresis (Orga Bio Human srl)

Experience and lab skills in molecular biology:

Nuclei isolation and nuclear extract preparation, RNA isolation and purification, Isolation of poly-A+ mRNA and synthesis of cDNA, RT-PCR cloning, Isolation and purification of plasmid and genomic DNA, Genotyping analysis, PCR-genotyping, Gene amplification, PCR and RT-PCR, Agarose and acrylamide gel electrophoresis, elution of DNA fragment from gel, Automated DNA sequencing, Southern blotting, Kinase end-labeling of DNA, Competent cell preparation, Restriction mapping of DNA, Recombinant DNA cloning, Primer labeling for primer extension assay, and Primer Extension, Northern blot, CGH array, Next Generation Sequencing

Computing Skills:

Applications: Microsoft Office Suite, Adobe suite, Statistical software;
Programming Languages: SQL, HTML, Office
Operating Systems: Unix, Windows

Teaching Skills:

ECM courses, Training courses of Lazio Region, University lessons in Genetic and molecular biology

Languages

Italian, English

RESEARCH ACTIVITY

2003 Identification of CF modifier genes by family studies and microarray analysis
2004 Pharmaceutical approaches for treatment of laminopathy
2005 Studi di associazione con numerosi polimorfismi di geni modificanti la funzione biochimico-strutturale del prodotto genico nella fibrosi cistica in pazienti genotipicamente omogenei; studio del ruolo dell'allele selvatico sulla neurodegenerazione nella malattia di Huntington
2006 Causes, evolution and progression of nasal polyps: role of modifier genes and a new approach through CGH array
2007 PROTHETS (Prognosis and therapeutic targets in the “Ewing” family of tumors)
2008 CHIP Array for the genomic analysis of microdeletions and microinsertions goldchip
2008 Identification of biomarkers for assessing the progression of cardiovascular disease in patient with Type 2 diabetes. Development of new kits for the measurement of NOX and circulating HMGB1
2010 Construction of a new computer system to support the early diagnosis of Alzheimer's dementia and in particular its pre-clinical condition known as Mild Cognitive Impairment
2010 Production molecules to inhibit the binding of ox-LDL with LOX-1 receptor for use in a therapeutic perspective
2010 Pharmacogenetics and pharmacogenomics of antiplatelet agents: development of innovative diagnostic kits
2011 Development of models and analysis systems in vivo, in vitro and in silico for the creation of drugs that inhibit receptor LOX-1, a new therapeutic target for cardiovascular disease.
2012 Pharmacogenetics and pharmacogenomics of antiplatelet agents: development of innovative diagnostic kits;
2013 Sviluppo di una piattaforma informatica di gestione/predizione del rischio genomico ed ambientale per le malattie cardiovascolari; TeMeGe (Tele Medicina Genomica)
2015 Role of DNA methylation in the risk of first and recurrent ischemic stroke
2015 Smad4 gene in patients with abdominal aortic aneurysm
2015 Identification of novel biomarkers for early diagnosis and monitoring disease course in Multiple Sclerosis
2015 Identification of molecular biomarkers for diagnosis, monitoring and prognosis of Parkinson and Alzheimer’s disease: new target for therapeutic approaches

CLINICAL and DIAGNOSTIC ACTIVITY

As Head of Laboratory of Molecular Genetic of Neuromed IRCCS, I have my expertise in Mendelian genetics, complex genetics, genomics,

and functional genomics with a primary focus on Neurodegenerative diseases. I carry out my diagnostic activity in cooperation with Alzheimer, Parkinson and Rare disease Units. The Laboratory of Genetic developed several diagnostic NGS panels for Spastic Paraplegia, Epilepsia, Emiplegic Migraine, Alzheimer Disease, Rare chanelopathies, Parkinson disease. We represent a regional excellence for Huntington, Spinocerebellar ataxias and a lot of different rare diseases.

TEACHING ACTIVITY

ECM Courses with Theta21, Rome (2011-2013)
ECM courses with Scuola Medica Ospedaliera, Rome (2010-Today)
Nutrire bene il tuo corpo alimenta la vita, Regione Lazio, 2011
Corso di Formazione Annuale della regione Molise "Giornalismo scientifico", sede IRCSS Neuromed, Pozzilli, Molise, Anno 2014

INVITED SPEAKER

28 October 2004, Toronto, Canada. Pyrosequencing ASHG (American Society of Human Genetics) Lunch Seminar: Pyrosequencing in Human Genetic Research. "Quantitative genotyping gives evidence of CAP70 as a possible modifier gene of Cystic Fibrosis Phenotype"
27 October 2006, Genova, Italy. Pancreatiti su base genetica: Inquadramento
6 June 2008, Rome, Italy. PROTHETS (Prognosis and therapeutic targets in the "Ewing" family of tumors): "Construction of an exon array CGH chip"
"Farmacogenetica ed applicazioni in oncologia". Aggiornamenti in oncologia. Nuovi aggiornamenti nella diagnosi e terapia. Aversa, 17 Dicembre 2014
La genetica molecolare nella pratica clinica. " Nuove strategie diagnostiche in genetica molecolare". Benevento, 7 Novembre 2013
Aggiornamento in oncologia 2015: nuovi orientamenti nella diagnosi e terapia. "Farmacogenetica ed applicazioni in oncologia" , 17 novembre 2015, Hotel del Sole, Aversa (Ce)
"La rivoluzione genomica implicazioni delle patologie cardiovascolari". Ciociaria cuore 2015, auditorium diocesano s. Paolo apostolo, Frosinone, 31 october 2015

SPEAKER IN TRAINING COURSE

"I test genetici ed il laboratorio di genetica. Dai test di nuova generazione alla certificazione: le nuove sfide" Scuola Medica Ospedaliera, Rome, 11 June 2012
"Genetica delle malattie Cardiovascolari", Artemisia Medical Group, Rome, 5 May 2012,
"miRNAe genetica: stato dell'arte", Istituto Mendel, Rome, 22 September 2012
"Le nuove frontiere della genetica: metodologie innovative e nuove applicazioni diagnostiche", Rome, from 5 May to 6 October 2012
"Test genetici: marcatura CE e patent", 21 November 2013 Scuola Medica Ospedaliera, Rome
"Laboratorio di genetica: dai test "classici" ai test di nuova generazione" ,10 June 2013, Scuola Medica Ospedaliera, Rome
"Patologie mendeliane nella diagnosi post e prenatale. Distrofinopatie e fibrosi cistica: iter diagnostici, interpretazione e gestione dei risultati", 25-26 March 2013, Scuola Medica Ospedaliera, Rome
"Genetica della malattia di Parkinson. La sfida diagnostica nelle neuroscienze: la genetica". 28 November 2014 IRCCS Neuromed
"Real Time PCR: Novità, orientamenti, percorsi diagnostici"; 10 June 2014, Istituto Mendel, Rome
"Dal clonaggio del gene ai nostri giorni: Mutazioni CFTR e forme atipiche"; 14 April 2014, Scuola Medica Ospedaliera, Roma
"Genetica del tumore alla mammella", 18 ottobre 2014 Giugliano
"Genetica del tumore alla mammella", 4 Ottobre 2014, Pozzuoli
"Genetica del tumore alla mammella". 6 Dicembre 2014
"Corso Strategie e Tecniche della Comunicazione Scientifica",2014 IRCCS Neuromed, Pozzilli, Isernia
"Malattie mendeliane: La fibrosi cistica. Dal clonaggio del gene ai nostri giorni". 14 Maggio 2015, Scuola Medica Ospedaliera, Rome
"Test genetici: marcatura CE e patent", 24 Settembre 2015, Scuola Medica Ospedaliera, Roma
"Genotipizzazione in Oncologia: Metodiche per la Valutazione dello stato RAS e prospettive future". New Drugs, New Acts: Mcrc - Neuromed Pozzilli, 4 dicembre 2015
"Farmacogenetica ed applicazioni in oncologia", Aggiornamenti in oncologia, Nuovi aggiornamenti nella diagnosi e terapia. Aversa, 17 Novembre 2015
"La rivoluzione genomica: implicazioni delle patologie cardiovascolari", Ciociaria Cuore, 31 Ottobre 2015 Frosinone
"Le nuove frontiere della genetica a sostegno delle malattier are", Giornata delle malattie rare 2016, 29 Febbraio 2016, Liceo Sceintifico A. Romita, Campobasso
"La familiarità del carcinoma mammario, rischio genetico ed ereditarietà"- Unite contro il tumore al seno; Avellino 22 Aprile 2016, Virginia palace hotel
Test genetici nelle CFTRpatie: utilità e limiti. Infertilità maschile e gene CFTR. Il "5T". Malattie correlate al gene cftr. Approcci diagnostici nella fibrosi cistica, 12 Maggio 2013, Istituto CSS-Mendel- Roma

REVIEWER FOR INTERNATIONAL JOURNALS

British Journal of Dermatology, The Laryngoscope, Genomic Insight, Gene Expression to Genetical Genomics, Journal of Pediatric

Genetics, Frontiers in Neuroesciense

MEMBER OF EDITORIAL BOARD
Journal of Clinical and Experimental Medicine, Genomic insight

SCIENTIFIC PAPER
H index: 8
Citations: 349

1. R. Ferese, R. Campopiano, V. Albano, L. Scorzolini, E. Giardina, S. Scala, C. D'Alessio, S. Zampatti, R. Fantozzi, M. Storto, G. Novelli and S. Gambardella. PCR based approach for molecular analysis of six neurotropic pathogens. Submitted to Acta Virologica

2. Lenzi P, Lazzeri G, Biagioni F, Busceti CL, Gambardella S, Salvetti A, Fornai F .The Autophagoproteasome a Novel Cell Clearing Organelle in Baseline and Stimulated Conditions.. Front Neuroanat. 2016 Jul 21;10:78.

3. M. Ferrucci, F. Biagioni, P. Lenzi, S. Gambardella, R. Ferese, M.T. Calierno, A. Falleni, A. Grimaldi, A. Frati, V. Esposito, C. Limatola, F. Fornai. Rapamycin promotes gene and protein expression related to cell differentiation while suppressing stemness and cell migration. Subbmitted to aging

4. S. Gambardella, R. Ferese, F. Biagioni, C. Busceti, R. Campopiano, A.M.P. Griguoli, G. Novelli, M. Storto and F. Fornai. Non Motor symptoms in genetic forms of Parkinsonism. Subbitted to Parkinson's Disease.

5. Gambardella S, Biagioni F, Ferese R, Busceti CL, Frati A, Novelli G, Ruggieri S and Fornai F (2016) Vacuolar Protein Sorting Genes in Parkinson's Disease: A Re-appraisal of Mutations Detection Rate and Neurobiology of Disease. Front. Neurosci. 10:532. doi: 10.3389/fnins.2016.00532

6. A New Splicing Mutation in the L1CAM Gene Responsible for X-Linked Hydrocephalus (HSAS). Ferese, R., Zampatti, S., Griguoli, A. M. P., Fornai, F., Giardina, E., Barrano, G., Albano, V., Campopiano, R., Scala, S., Novelli, G. & Gambardella, S. Journal of Molecular Neuroscience. 2016 p. 1-6 6 p.

7. Four Copies of SNCA Responsible for Autosomal Dominant Parkinson's Disease in Two Italian Siblings. Ferese, R., Modugno, N., Campopiano, R., Santilli, M., Zampatti, S., Giardina, E., Nardone, A., Postorivo, D., Fornai, F., Novelli, G., Romoli, E., Ruggieri, S. & Gambardella, S. Parkinson's Disease. 2015, 546462

8. Direct PCR: A new pharmacogenetic approach for the inexpensive testing of HLA-B∗57:01. R. Cascella; C. Strafella; M. Ragazzo; S. Zampatti; P. Borgiani; S. Gambardella; A. Pirazzoli; G. Novelli; E. Giardina. Pharmacogenomics Journal. 2015;15(2):196-200.

9. Short history of the "Genomic Revolution" and implication for neurological institutes. Stefano Gambardella; Veronica Albano; Rosa Campopiano; Rosangela Ferese; Simona Scala; Marianna Storto; Stefania Zampatti; Edoardo Romoli; Rivista Italiana della Medicina di Laboratorio. 2015;11(1).

10. Novel mutations of the TCOF1 gene in European patients with Treacher Collins syndrome. Conte C. D'Apice MR. Rinaldi F. Gambardella S. Sangiuolo F. Novelli G. 1471-2350 12 1 2011 BMC medical genetics

11. The etiology of acute recurrent pancreatitis in children: a challenge for pediatricians. Lucidi V. Alghisi F. Dall'Oglio L. D'Apice MR. Monti L. De Angelis P. Gambardella S. Angioni A. Novelli G. 1536-4828 40 4 2011 May Pancreas

12. Bone marrow and umbilical cord blood human mesenchymal stem cells: state of the art. Malgieri A. Kantzari E. Patrizi MP. Gambardella S. 1940-5901 3 4 2010 Int J Clin Exp Med

13. New PRSS1 and common CFTR mutations in a child with acute recurrent pancreatitis, could be considered an "Hereditary" form of pancreatitis ? Corleto VD. Gambardella S. Gullotta F. D'Apice MR. Piciucchi M. Galli E. Lucidi V. Novelli G. Delle Fave G. 1471-230X 10 2010 BMC Gastroenterology

14. A fluorescence-based sequence-specific primer PCR for the screening of HLA-B(*)57:01. Giardina E. Stocchi L. Foti Cuzzola V. Zampatti S. Gambardella S. Patrizi MP. Bramanti P. Pirazzoli A. Novelli G. 1522-2683 31 21 2010 Oct Electrophoresis

15. Overexpression of microRNA-206 in the skeletal muscle from myotonic dystrophy type 1 patients. Gambardella S. Rinaldi F. Lepore SM. Viola A. Loro E. Angelini C. Vergani L. Novelli G. Botta A. 1479-5876 8 2010 J Transl Med

16. Variations of inflammatory mediators and alpha1-antitrypsin levels after lung volume reduction surgery for emphysema. Mineo D.

Ambrogi V. Cufari ME. Gambardella S. Pignotti L. Pompeo E. Mineo TC. 1535-4970 181 8 2010 Apr 15 Am. J. Respir. Crit. Care Med.

17. Design, Construction and Validation of Targeted BAC Array-Based CGH Test for Detecting the Most Commons Chromosomal Abnormalities. S.Gambardella, E. Ciabattoni, F. Motta, G.Stoico, F. Gullotta, M. Biancolella, AM. Nardone, A. Novelli, E. Brunetti, L. Bernardini and G. Novelli. 2010:3 9-21, 11 Mar 2010 Genomics Insights

18. Hyperproliferation in nasal polyposis tissues is not associated with somatic genomic instability. Corradini C. Gullotta F. Ciacci S. Palmieri G. Salehi LB. De Corso E. Novelli G. Gambardella S. 1916-0216 37 4 2008 Aug Journal of otolaryngology - head & neck surgery

19. Phenotypic variability in a family with pancreatitis and cystic fibrosis sharing common mild CFTR mutation: report on CFTR mutations and their phenotypic variability. Alghisi F. Bella S. Lucidi V. Angioni A. Tomaiuolo AC. D'Apice MR. Gambardella S. Novelli G. 1536-4828 38 1 2009 Jan Pancreas

20. Screening of EDA1 gene in X-linked anhidrotic ectodermal dysplasia using DHPLC: identification of 14 novel mutations in Italian patients. Conte C. Gambardella S. Bulli C. Rinaldi F. Di Marino D. Falconi M. Bramanti P. Desideri A. Novelli G. 1090-6576 12 3 2008 Sep Genet. Test.

21. Gene symbol: ED1. Disease: X-linked anhidrotic ectodermal dysplasia. Gambardella S. 1432-1203 123 1 2008 Feb Hum. Genet.

22. Gene symbol: ED1. Disease: Ectodermal dysplasia. Gambardella S. 1432-1203 123 1 2008 Feb Hum. Genet.

23. Gene symbol: ED1. Disease: Ectodermal dysplasia. Gambardella S. 1432-1203 123 1 2008 Feb Hum. Genet.

24. Gene symbol: ED1. Disease: Ectodermal dysplasia. Gambardella S. 1432-1203 123 1 2008 Feb Hum. Genet.

25. Gene expression analysis in myotonic dystrophy: indications for a common molecular pathogenic pathway in DM1 and DM2. Botta A. Vallo L. Rinaldi F. Bonifazi E. Amati F. Biancolella M. Gambardella S. Mancinelli E. Angelini C. Meola G. Novelli G. 1052-2166 13 6 2007 Gene Expr.

26. Denaturing HPLC in laboratory diagnosis of hereditary angioedema. Guarino MD. Perricone C. Guarino S. Gambardella S. D'Apice MR. Fontana L. Novelli G. Perricone R. 0091-6749 120 4 2007 Oct J. Allergy Clin. Immunol.

27. Gene expression profile study in CFTR mutated bronchial cell lines. Gambardella S. Biancolella M. D'Apice MR. Amati F. Sangiuolo F. Farcomeni A. Chillemi G. Bueno S. Desideri A. Novelli G. 1591-8890 6 4 2006 Dec Clin. Exp. Med.

28. Gonadal mosaicism in hereditary angioedema. Guarino S. Perricone C. Guarino MD. Giardina E. Gambardella S. Rosaria D'Apice M. Bulli C. Perricone R. Novelli G. 0009-9163 70 1 2006 Jul Clin. Genet.

29. Segregation analysis in cystic fibrosis at-risk family demonstrates that the M348K CFTR mutation is a rare innocuous polymorphism. D'Apice MR. Gambardella S. Russo S. Lucidi V. Nardone AM. Pietropolli A. Novelli G. 0197-3851 24 12 2004 Dec 15 Prenat. Diagn.

30. Gene expression profiling of fibroblasts from a human progeroid disease (mandibuloacral dysplasia, MAD #248370) through cDNA microarrays. Amati F. Biancolella M. D'Apice MR. Gambardella S. Mango R. Sbraccia P. D'Adamo M. Margiotti K. Nardone A. Lewis M. Novelli G. 1052-2166 12 1 2004 Gene Expr.

31. Toward the pharmacogenomics of cystic fibrosis--an update. Sangiuolo F. D'Apice MR. Gambardella S. Di Daniele N. Novelli G. 1462-2416 5 7 2004 Oct Pharmacogenomics

32. Molecular analysis using DHPLC of cystic fibrosis: increase of the mutation detection rate among the affected population in Central Italy. D'Apice MR. Gambardella S. Bengala M. Russo S. Nardone AM. Lucidi V. Sangiuolo F. Novelli G. 1471-2350 5 2004 Apr 14 BMC Med. Genet.

33. Utilizzo della metodica DHPLC per lo screening delle mutazioni del gene CFTR nella diagnosi molecolare di Fibrosi Cistica.S. Gambardella, M.R. D'Apice, S. Russo, M. Bengala, A.M. Nardone, V. Lucidi, F. Sangiuolo, G. Novelli. Biologi Italiani Maggio 2003

ABSTRACT FOR PROFESSIONAL CONFERENCE AND MEETING

Screening allelico CFTR nella popolazione italiana mediante DHPLC. M.R. D'Apice, F. Sangiuolo, **S. Gambardella**, S. Russo, M. Lais, A. Ranciaro, G. Novelli. *5th Meeting Nazionale della Società Italiana di Genetica Umana (S.I.G.U.)*, Verona, Italy, 24-27 September 2002.

CAP70 as a possible modifier gene of Cystic fibrosis phenotype. **Gambardella S**. et al. *54th Annual Meeting of American Society of Human Genetics*, Toronto, Canada October 26 – 30 2004.

CAP70: nuovo gene modificatore del fenotipo nella Fibrosi Cistica? **Gambardella S**. et al. 7° Congresso Nazionale S.I.G.U. 2004.

Different CFTR genotypes induce dissimilar expression patterns in CF putative modifier genes. **Gambardella S**. et al. *European Human Genetics Conference* 2005, Prague 7-10 May 2005.

Different CFTR genotypes induce dissimilar expression patterns in CF putative modifier genes. **Gambardella S**. et al. *28th European CF Conference*, Crete, Greece 22-25 June 2005.

Differenti genotipi CFTR causano un diverso adattamento dell'espressione genica in linee cellulari FC. **Gambardella S.** et al. *VIII Congresso Nazionale S.I.G.U*. 28-30 settembre 2005.

Gene expression analysis in Myotonic Dystrophy: Indications for a common molecular pathogenic pathway in DM1 and DM2. Rinaldi F, Botta A, Vallo L, Bonifazi E, **Gambardella S**, Mancinelli E, Angelini C, Meola G, Novelli G. *IDMC-6, 6th International Myotonic Distrophy Consortium Meeting*, Milano 12-15 September 2007.

A case of incomplete penetrance in two carriers of CFTR and PRSS1 mutations; Digestive and Liver Disease S. Ciacci, **S. Gambardella**, M.R. D'Apice, S. Petrocchi, V. Lucidi, M. Bengala, G. Novelli. Chronic pancreatitis:, *Volume 39, Issue 10, Pages A56-A57 Published on 2007-31-10*

Recurrent pancreatitis as the first manifestation of cystic fibrosis: a single centre experience. V. Lucidi, F. Alghisi, A. Angioni, A.C. Tomaiuolo, M.R. D'Apice, **S. Gambardella**, B. Russo, S. Bella, P. De Angelis, L. Dall'Oglio, G. Novelli. *Journal of Cystic Fibrosis, Volume 7, Supplement 3,Pages S19-S20 Published on 2008-31-07*

Phenotypic variability in a family with pancreatitis and cystic fibrosis sharing common mild CFTR mutation. V. Lucidi, F. Alghisi, A. Angioni, A.C. Tomaiuolo, M.R. D'Apice, **S. Gambardella**, B. Russo, S. Bella, E. Fiscarelli, G. Novelli. *Journal of Cystic Fibrosis, Volume 7, Supplement 3,Page S10 Published on 2008-31-07*

GOLD (Gain or Loss Detection) Chip: a new array CGH tool for genetic diagnosis. S. Comuzio, E. Ciabattoni, F. Motta, G. Stoico, E. Bartolini, G. Babbini, A. Novelli, G. Novelli, F. Gullotta, **S. Gambardella.** *58th Annual Meeting of American Society of Human Genetics*, Philadelpia, USA, November 11-15, 2008.

A New Compound Heterozygous Mutation of PRSS1 And CFTR Genes as Cause of Recurrent Acute Pancreatitis. Piciucchi M, Corleto VD, **Gambardella S**, Gullotta F, D'Apice MR, Lucidi V, Novelli G, Delle Fave G. *AISP - 32nd National Congress.* Montecatini Terme, PT (Italy). October 2-4, 2008, JOP. J Pancreas (Online) 2008; 9(6 Suppl):844-845.

Potenzialità di ricerca su liquido amniotico: DNA microarray (il gold chip). G. Stoico, **S. Gambardella**, E. Ciabattoni, F. Motta, L. Ferretti, E. Bartolini, A. Novelli, F. Gullotta, S. Comuzio, G. Novelli. *ATTI della Società Italiana di Ginecologia e Ostetricia - Vol. LXXXV*

Scienz!: il network scientifico integrato. **S. Gambardella**, P. Rosati, A. Dell'Anna, R. Marini, S. Comuzio, S. De Rosa, M. Savona, G. Novelli. *Società Italiana di Genetica Umana*, Firenze 14-17 Ottobre 2010

A new potential and powerful tool for prenatal diagnosis: GOLDchip. G. Stoico, **S. Gambardella**, E. Ciabattoni, L. Ferretti, S. Piccininni, E. Bartolini, A. Novelli, A. Nardone, S. Comuzio, G. Novelli. 2° *Congress of the European Society of Predictive* Medicine Berlin, 14-15 May 2011

Sordità congenita: sviluppo di un rapido ed innovativo saggio molecolare per l'indagine delle varianti 35delG e M34T nel gene GJB2 R. Cascella, C. Peconi, S. Zampatti, L. Stocchi, **S. Gambardella**, P. Patrizi, E. Giardina, G. Novelli; *Società Italiana di Genetica Umana*, Milano 13-16 novembre 2011

Protocollo diagnostico per la caratterizzazione molecolare della Distrofia muscolare di Duchenne (DMD) e di Becker (BMD) attraverso multiplex PCR ed elettroforesi capillare. **S. Gambardella**, , R. Marini,, R. Cascella, G. Barrano, M. Di Gregorio, A. Sabino, A.M. Nardone, A. Mesoraca, E. Giardina, G. Galluzzi, L. Seminara, G. Sabbadini. *Società Italiana di Genetica Umana*, Sorrento 21-23 Novembre 2012

A-synuclein triplication inside a duplicated region in an Italian siblings with Parkinson disease. R. Ferese, M. Santilli, R. Campopiano, N. Modugno,V. Albano, S. Scala, S. Gambardella, S. Zampatti, M. Storto, E. Giardina, A. Nardone, D. Postorivo, F. Fornai, G. Novelli, S. Ruggieri, E. Romoli. 28 Congresso Nazionale della Società Italiana di Medicina di Laboratorio (SIPMEL), Rimini
29-30 Ottobre 2014

Sviluppo di protocolli diagnostici innovativi per la genotipizzazione di Varianti genomiche clinicamente associate alle patologie Cardiovascolari. S. Scala, D. Rosati, S. Gambardella, R. Campopiano, V. Albano, R. Ferese, M. Storto, E. Romoli. 28 Congresso Nazionale della Società Italiana di Medicina di Laboratorio (SIPMEL), Rimini 29-30 Ottobre 2014

Use of in-house protocols for neurotropic virus detection in Molecular routine: cheap analysis with a good sensibility and specificity. S. Gambardella, R. Ferese, V. Albano, S. Scala, R. Campopiano, M. Storto, M. Leva, E. Romoli 28 Congresso Nazionale della Società Italiana di Medicina di Laboratorio (SIPMEL), Rimini 29-30 Ottobre 2014

Four Copies Of SNCA Responsible Of Autosomal Dominant Parkinson's Disease In Two Italian Siblings. The International Parkinson and Movement Disorder 2015, San Diego, California, USA June 14-18, 2015. MDS 19th International Congress of Parkinson's Disease and Movement Disorders. Volume 30, June 2015 Abstract, Volume 30, June 2015 Abstract Supplement

ADVANCED COURSES
5-12 November 2006, Rome
First ROC international workshop and practical course on chromatin immunoprecipitation related techniques.

Autorizzo il trattamento dei dati in conformità a quanto previsto dal DL 196/03.

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