Beatrice Spedicati

Date of birth: 04/07/1992 | **Nationality:** Italian | **Gender:** Female

WORK EXPERIENCE

I.R.C.C.S. "BURLO GAROFOLO" - S.C. GENETICA MEDICA – TRIESTE, ITALY

ATTENDING PHYSICIAN - MEDICAL GENETICIST – 15/11/2023 – CURRENT

Her diagnostic activity comprises clinical consultancies for patients affected by genetic conditions in the prenatal,neonatal, and postnatal settings, with a specific focus on critically ill newborns and patients affected by hearing loss, hereditary cardiovascular disorders, and hereditary ocular diseases. She also performs Next Generation Sequencing data interpretation (targeted-resequencing panels, whole exome sequencing and whole genome sequencing) in a multidisciplinary team that includes Biologists and Bioinformaticians. Additionally, she is the coordinator of the Medical Genetics team of the I.R.C.C.S. “Burlo Garofolo” that takes part in the Italian Register for the Clinical and Scientific Research on Pitt-Hopkins Syndrome.

DEPARTMENT OF MEDICINE, SURGERY AND HEALTH SCIENCES, UNIVERSITY OF TRIESTE – TRIESTE, ITALY

RESEARCHER (RTDB) – 02/10/2023 – CURRENT

Her research work is focused on the genetic of senses and is aimed at identifying genes involved both in physiological sensory functions and in monogenic and complex disorders of the sensory systems. Furthermore, she has cooperated in the definition of the Italian guidelines for the genetic analysis of hereditary hearing loss (document currently under revision by the Società Italiana di Genetica Umana - SIGU). She is also collaborating with several international consortia as CKDgen consortium, Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) consortium, and ThyroidOmics Consortium. Additionally, she is the vice-coordinator of the Medical Genetics team of the I.R.C.C.S. “Burlo Garofolo” that takes part into the national Rete IDEA Research project on undiagnosed patients and she is one of the main contributors to the “Iconodiagnosis” project developed by the Medical Genetics team of the I.R.C.C.S. “Burlo Garofolo” in collaboration with several Italian geneticists and pediatricians.

I.R.C.C.S. "BURLO GAROFOLO" – TRIESTE, ITALY

MASTER SCHOLARSHIP – 01/02/2023 – 30/09/2023

5x1000 2015 Project "Senses - Genetics of senses and related diseases"; CUP:C92F17003560001.

EDUCATION AND TRAINING

01/11/2021 – 21/03/2025 Trieste, Italy

PHD PROGRAMME IN REPRODUCTION AND DEVELOPMENTAL SCIENCES Department of Medicine, Surgery and Health Sciences, University of Trieste

She obtained the European label for her doctoral degree, as per initiative of the European University Association, as she was able to fulfill all four requirements for its achievement (i.e. joint supervision, an international Board of Examiners, multilingualism and international mobility).
During her PhD programme, she participated in several research projects focusing primarily on the genetic of senses, i.e. hearing, smell, taste and sight. In particular, she was involved in the study of the genetic bases of normal hearing function, monogenic and multifactorial hearing loss, smell function and dysfunction (especially in correlation with aging and, recently, with SARS-CoV-2 infection), taste, food preferences, and colour vision defects. She also took part in other research projects, as the identification of the genetic bases of endometriosis and the genetics of odontostomatological traits. Additionally, she expanded her research projects on the clinical and genetic characterisation of “Human Knockouts” and on dual molecular diagnoses in complex patients.

Address Strada di Fiume, 447, 34149, Trieste, Italy | **Website** https://dsm.units.it/

01/11/2018 – 30/10/2022 Trieste, Italy

SPECIALISATION SCHOOL IN MEDICAL GENETICS Department of Medicine, Surgery and Health Sciences, University of Trieste

During the Specialisation School in Medical Genetics, she participated in more than 1500 clinical consultancies in the prenatal, neonatal, and postnatal settings, acquiring specific training in patients evaluation, diagnosis of genetic conditions, health-care management and available medical treatments. Furthermore, she was trained in Next Generation Sequencing data production and interpretation (targeted-resequencing panels, whole exome sequencing and whole genome sequencing). Along with the peculiar hard skills of the discipline, she also implemented her soft skills, as the ability to establish an efficient communication with patients, work in a multidisciplinary team, and apply an efficient problem-solving strategy.

Concerning her research activity, during her Specialisation School, she had the opportunity to contribute to the clinical and genetic characterisation of "Human Knockouts" in genetically isolated populations belonging to the INGI Consortium (i.e. Friuli-Venezia Giulia Genetic Park, Carlantino, Val Borbera) and to expand the research project on dual molecular diagnoses in complex patients.

Address Strada di Fiume, 447, 34149, Trieste, Italy | **Website** https://dsm.units.it/ | **Final grade** 50/50 cum laude |

Thesis Behind the scenes of complex models of inheritance: dual molecular diagnoses explain entangled clinical pictures

11/2017 – 02/2018 Trieste, Italy
STATE EXAM - PROFESSIONAL REGISTER ADMMISSION Department of Medicine, Surgery and Health Sciences, University of Trieste

Address Strada di Fiume, 447, 34149, Trieste, Italy | **Website** https://dsm.units.it/

10/2011 – 10/2017 Trieste, Italy
MASTER'S DEGREE IN MEDICINE AND SURGERY Department of Medicine, Surgery and Health Sciences, University of Trieste

Address Strada di Fiume, 447, 34149, Trieste, Italy | **Website** https://dsm.units.it/ | **Final grade** 110/110 cum laude |

Thesis Screening of the secretome to identify factors that promote cardiomyocyte proliferation

07/2016 – 10/2017 Trieste, Italy
MASTER'S DEGREE INTERNSHIP International Centre for Genetic Engineering and Biotechnology - Molecular Medicine Group

- DNA, RNA and protein extraction
- Gene expression analysis using Real-time PCR
- Western Blotting analysis
- Bacterial transformation
- Plasmid amplification and purification
- RNA interference
- Primary mammalian cells isolation and culture
- Cells transfection and infection procedures
- Immunofluorescence assays on cells and tissues

Address AREA Science Park, Padriciano, 99, 34149, Trieste, Italy | **Website** https://www.icgeb.org/

11/2004 – 06/2016 Trieste, Italy
DEGREE IN ORGAN AND ORGAN COMPOSITION "G. Tartini" State Conservatory of Trieste

Address Via Ghega, 12, 34132, Trieste, Italy | **Website** https://conts.it/ | **Final grade** 8/10

09/2006 – 07/2011 Trieste, Italy
SECONDARY SCHOOL DIPLOMA "G. Oberdan" Scientific High School

Address Via Veronese, 1, 34144, Trieste, Italy | **Website** https://www.liceo-oberdan.edu.it/ | **Final grade** 100/100

NATIONAL AND INTERNATIONAL CONFERENCES AND SEMINARS - ORAL PRESENTATIONS

13/11/2019 – 16/11/2019
XXII SIGU National Congress (Rome, Italy)

"We are all experiments of nature: the fascinating role of Human Knockouts"
Authors:
B. Spedicati, R. Palmisano, M. Cocca, C. Barbieri, F. Sirchia, M. Mezzavilla, A. Morgan, F. Faletra, P. Gasparini, G. Girotto

Link https://sigu.congressonazionale.com/congressi-precedenti/

22/03/2021 – 24/03/2021
Genomics of Rare Disease (Online conference)

"Natural Human Knockouts and deep phenotyping in Italian genetic isolates: a different perspective on autosomal recessive Mendelian disorders"
Authors:
B. Spedicati, M. Cocca, R. Palmisano, F. Faletra, C. Barbieri, M. Francescatto, M. Mezzavilla, A. Morgan, G. Pelliccione, P. Gasparini, G. Girotto

Link https://coursesandconferences.wellcomeconnectingscience.org/event/genomics-of-rare-disease-virtualconference- 20210322/

10/09/2022 – 13/09/2022
Symposium & 57th Inner Ear Biology Workshop – IEB 2022 (Trieste, Italy)

"Dual molecular diagnosis in complex hearing loss patients: when a single gene is not enough"
Authors:

B. Spedicati, A. Morgan, U. Ambrosetti, L. Garavelli, S. Lenarduzzi, G. Pelliccione, F. Peluso, A. Santin, P. Gasparini, G. Girotto

Link https://ieb2022.it/

04/10/2023 – 06/10/2023
XXVI SIGU National Congress (Rimini, Italy)

"Empowering the application of the molecular autopsy in Sudden Cardiac Deaths (SCD): the experience of the Friuli-Venezia Giulia (FVG) Regional Register"
Authors:
B. Spedicati, S. Lenarduzzi, M. Dal Ferro, A. Paldino, D. Mazzà, M. Zecchin, S. D'Errico, G. Sinagra, P. Gasparini, G. Girotto

08/04/2024 – 10/04/2024
Human Genome Meeting (HGM) 2024 (Rome, Italy)

"When time matters: application of high-throughput sequencing technologies in critically-ill infants admitted to neonatal and pediatric intensive care units (NICU/PICU)"
Authors:
B. Spedicati, L. Memo, A. Morgan, S. Zampieri, E. Paccagnella, G. Zucca, M. Troian, E. Agolini, A. Dotta, E. Rosina, L. Pezzani, A. Novelli, M. Iascone, G. Girotto

02/10/2024 – 04/10/2024
XXVII SIGU National Congress (Padova, Italy)

"A race against time: when high-throughput sequencing technologies are essential to provide critically-ill children admitted to Neonatal and Pediatric Intensive Care Units (NICU/PICU) with a molecular diagnosis"
Authors:
B. Spedicati, L. Memo, A. Morgan, S. Zampieri, E. Paccagnella, F. Perino, S. Frisari, M. Troian, E. Agolini, A. Dotta, E. Rosina, C. Lucca, L. Pezzoli, L. Pezzani, A. Novelli, M. Iascone, G. Girotto

Link https://sigu.congressonazionale.com/

NATIONAL AND INTERNATIONAL CONFERENCES AND SEMINARS - POSTER PRESENTATIONS

06/06/2020 – 09/06/2020
European Human Genetics Conference (Virtual conference)

"The importance of Human Knockouts in a deeper characterization of Mendelian disorders"
Authors:
B. Spedicati, F. Faletra, R. Palmisano, C. Barbieri, G. Pelliccione, A. Morgan, M. Mezzavilla, M. Cocca, P. Gasparini, G. Girotto

Link https://2020.eshg.org/

11/11/2020 – 13/11/2020
XXIII SIGU National Congress (Virtual conference)

"When mosaicism deceives the eye: an incidental diagnosis of Beckwith-Wiedemann syndrome"
Authors:
B. Spedicati, B. Bosio, A.P. D'Adamo, S. Cappellani, A. Feresin, P. Gasparini, G. Girotto, F. Faletra, C. Ardisia.

Link https://sigu.congressonazionale.com/

28/08/2021 – 31/08/2021
European Human Genetics Conference (Virtual conference)

"There's more than meets the eye: dual molecular diagnosis in complex hearing loss patients"
Authors:
B. Spedicati, A. Morgan, M. Bonati, G. Severi, A. Feresin, G. Pelliccione, P. Tesolin, C. Graziano, P. Gasparini, F. Faletra, G. Girotto

Link https://2021.eshg.org/

17/11/2021 – 19/11/2021
XXIV SIGU National Congress (Virtual conference)

"Whole Exome Sequencing in the pediatric emergency setting: when time matters for patients' treatment, care and management"
Authors:
B. Spedicati, A. Feresin, L. Musante, A. Morgan, M. La Bianca, F. Faletra, M.T. Bonati, E. Rubinato, G. Girotto, P. Gasparini

Link https://sigu.congressonazionale.com/2021/

11/06/2022 – 14/06/2022
European Human Genetics Conference (Hybrid conference Vienna - Virtual)

"Persistent chemosensory dysfunction in COVID-19 patients: a deep dive into the psychophysical and genetic characterisation of an Italian cohort"
Authors:
B. Spedicati, G. G. Nardone, A. Santin, A. Morgan, M. P. Concas, G. Tirelli, P. Gasparini, P. Boscolo-Rizzo, G.Girotto

Link https://2022.eshg.org/

07/09/2022 – 09/09/2022
XXV SIGU National Congress (Trieste, Italy)

"Behind the scenes of entangled clinical pictures: the intriguing role of dual molecular diagnoses"
Authors:
B. Spedicati, A. Morgan, M.T. Bonati, A. Luglio, E. Rubinato, S. Suergiu, P. Gasparini, F. Faletra, G. Girotto

Link https://sigu.congressonazionale.com/2022/

07/09/2022 – 09/09/2022
XXV SIGU National Congress (Trieste, Italy)

"Unravelling the genetic bases of persistent olfactory dysfunction in COVID-19 patients: the psychophysical and molecular characterisation of a large Italian cohort"
Authors:
B. Spedicati, G.G. Nardone, M.P. Concas, F. Crudele, A. Pecori, A. Santin, G. Tirelli, P. Gasparini, A. Morgan, P. Boscolo-Rizzo, G. Girotto

Link https://sigu.congressonazionale.com/2022/

10/09/2022 – 13/09/2022
Symposium & 57th Inner Ear Biology Workshop – IEB 2022 (Trieste, Italy)

"There's more behind Hereditary Hearing Loss: molecular and phenotypic expansion of *PPP1R12A*-related disorder"
Authors:
B. Spedicati, A. Morgan, L. Garavelli, G. G. Nardone, G. Pelliccione, G. Pianigiani, P. Gasparini, G. Girotto

Link https://ieb2022.it/

10/06/2023 – 13/06/2023
European Human Genetics Conference (Hybrid conference Glasgow - Virtual)

"Unveiling the genetic bases of Hereditary Hearin Loss (HHL): the application of a multistep diagnostic approach in a large Italian cohort"
Authors:
B. Spedicati, A. Santin, G. G. Nardone, S. Lenarduzzi, E. Rubinato, C. Graziano, L. Garavelli, S. Miccoli, S. Bigoni, A. Morgan, G. Girotto

Link https://2023.eshg.org/

02/09/2023 – 05/09/2023
Symposium & 58 Inner Ear Biology Workshop – IEB 2023 (London, UK)

"Unravelling the Genetic Bases of Hearing Loss: a Multistep and Integrative Approach in a Deeply Characterised Italian Cohort"
Authors:
B. Spedicati, A. Santin, G.G. Nardone, S. Lenarduzzi, E. Paccagnella, E. Rubinato, A. Morgan, G. Girotto

04/10/2023 – 06/10/2023
XXVI SIGU National Congress (Rimini, Italy)

"Rapid Whole Exome Sequencing (rWES) analysis in critically-ill newborns: when a precise and early diagnosis is fundamental for prompt and tailored management"
Authors:
B. Spedicati, E. Paccagnella, E. Rubinato, A. Salvador, G. Pelliccione, A. Morgan, G. Girotto

01/11/2023 – 05/11/2023
American Society of Human Genetics Annual Meeting 2023 (Washington DC, USA)

"Puzzling out the genetic bases of hereditary cardiovascular diseases: application of an integrative approach in a deeply clinically characterised Italian cohort"
Authors:
B. Spedicati, S. Lenarduzzi, M. Dal Ferro, A. Paldino, D. Mazzà, S. D'Errico, G. Sinagra, P. Gasparini, G. Girotto

01/06/2024 – 04/06/2024
European Human Genetics Conference (Hybrid conference Berlin - Virtual)

"he diagnostic chameleon of Hereditary Hearing Loss (HHL): a genetic investigation on Non-syndromic Mimics (NSMs)"
Authors:
B. Spedicati, A. Morgan, S. Lenarduzzi, E. Rubinato, G. Zucca, M. Troian, D. Marangoni, G. Girotto

02/10/2024 – 04/10/2024
XXVII SIGU National Congress (Padova, Italy)

"Unmasking Non-Syndromic Mimics (NSMs) in Hereditary Hearing Loss (HHL) patients: a genetic and clinical investigation in the Italian population"
Authors:
B. Spedicati, S. Lenarduzzi, A. Morgan, G. Pianigiani, C. Bon, E. Rubinato, D. Marangoni, G. Girotto

24/05/2025 – 27/05/2025
European Human Genetics Conference (Hybrid conference Milan - Virtual)

“When timing is essential: application of next-generation sequencing (NGS) technologies to provide a molecular diagnosis for critically-ill children admitted to Neonatal and Pediatric Intensive Care Units (NICUs/PICUs)”.
Authors:
B. Spedicati, L. Memo, A. Morgan, S. Zampieri, E. Paccagnella, F. Perino, S. Frisari, M. Troian, E. Agolini, A. Dotta, E. Rosina, C. Lucca, L. Pezzoli, L. Pezzani, A. Novelli, M. Iascone, G. Girotto

23/09/2025 – 25/09/2025
XXVIII SIGU National Congress (Rimini, Italy)

"Do not judge by the appearances: Whole Exome Sequencing (WES) reveals the presence of Non-Syndromic Mimics (NSMs) in Hereditary Hearing Loss (HHL) patients".
Authors:
B. Spedicati, G. Pianigiani, S. Lenarduzzi, E. Rubinato, A. Morgan, G. Girotto

14/10/2025 – 18/10/2025
American Society of Human Genetics Annual Meeting 2023 (Boston, USA)

"Running against the clock: application of Next Generation Sequencing (NGS) technologies to diagnose critically-ill children admitted to Neonatal and Pediatric Intensive Care Units (NICUs/PICUs)".
Authors:
B. Spedicati, L. Memo, A. Morgan, S. Zampieri, E. Paccagnella, S. Frisari, E. Agolini, A. Dotta, E. Rosina, C. Lucca, L. Pezzoli, L. Pezzani, A. Novelli, M. Iascone, G. Girotto

PUBLICATIONS

2020
Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population

Authors:
Anna Morgan, Stefania Lenarduzzi, **Beatrice Spedicati**, Elisabetta Cattaruzzi, Flora Maria Murru, Giulia Pelliccione, Daniela Mazzà, Marcella Zollino, Claudio Graziano, Umberto Ambrosetti, Marco Seri, Flavio Faletra, Giorgia Girotto

https://doi.org/10.3390/genes11111237

2020
COVID-19 experience: first Italian survey on healthcare staff members from a Mother-Child Research Hospital using combined molecular and rapid immunoassays test

Authors:
Manola Comar, Marco Brumat, Maria Pina Concas, et al. (including **Beatrice Spedicati**)
medRxiv preprint

doi.org/10.1101/2020.04.19.20071563

2021
The Role of Knockout Olfactory Receptor Genes in Odor Discrimination

Authors:
Maria Pina Concas, Massimiliano Cocca, Margherita Francescatto, Thomas Battistuzzi, **Beatrice Spedicati**, Agnese Feresin, Anna Morgan, Paolo Gasparini and Giorgia Girotto

https://doi.org/10.3390/genes12050631

2021
TBC1D24 and non-syndromic autosomal dominant hearing loss: the identification of an additional Italo-American family carrying the p.(S178L) mutation

Authors:
Beatrice Spedicati, Anna Morgan, Flavio Faletra, Agnese Feresin,Giulia Pelliccione, Paolo Gasparini, Giorgia Girotto

2021
Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates

Authors:
Beatrice Spedicati*, Massimiliano Cocca*, Roberto Palmisano, Flavio Faletra, Caterina Barbieri, Margherita Francescatto, Massimo Mezzavilla, Anna Morgan, Giulia Pelliccione, Paolo Gasparini, Giorgia Girotto
* B.S. and M.C. are joined first authors

https://doi.org/10.1038/s41431-021-00850-9

2022
Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals

Authors:
Thomas W. Winkler, Humaira Rasheed, Alezander Teummer, et al. (including **Beatrice Spedicati**)

https://doi.org/10.1038/s42003-022-03448-z

2022
Infant with a big head and 'crossed' polysyndactyly

Authors:
Gianluca Tamaro, Francesco Baldo, **Beatrice Spedicati**, Andrea Taddio, Flavio Faletra, Egidio Barbi

https://doi.org/10.1111/jpc.16063

2022
Challenging Occam's Razor: Dual Molecular Diagnoses Explain Entangled Clinical Pictures

Authors:
Beatrice Spedicati, Anna Morgan, Giulia Pianigiani, Luciana Musante, Elisa Rubinato, Aurora Santin, Giuseppe Giovanni Nardone, Flavio Faletra and Giorgia Girotto

https://doi.org/10.3390/genes13112023

2022
Impact of cultural and genetic structure on food choices along the Silk Road

Authors:
Serena Aneli, Massimo Mezzavilla, Eugenio Bortolini, Nicola Pirastu, Giorgia Girotto, **Beatrice Spedicati**, Paola Berchialla, Paolo Gasparini, and Luca Pagani

https://doi.org/10.1073/pnas.2209311119

2022
Exome sequencing efficacy and phenotypic expansions involving esophageal atresia/tracheoesophageal fistula plus

Authors:
Mary R. Sy, Jaynee Chauhan, Katrina Prescott et al. (including **Beatrice Spedicati**)

https://doi.org/10.1002/ajmg.a.62976

2023
The enigmatic genetic landscape of Hereditary Hearing Loss: a multistep diagnostic strategy in the Italian population

Authors:
Beatrice Spedicati, Aurora Santin, Giuseppe Giovanni Nardone, Elisa Rubinato, Stefania Lenarduzzi, Claudio Graziano, Livia Garavelli, Sara Miccoli, Stefania Bigoni, Anna Morgan, Giorgia Girotto

https://doi.org/10.3390/biomedicines11030703

2023
Whole-exome sequencing: Clinical characterization of pediatric and adult Italian patients affected by different forms of hereditary cardiovascular diseases

Authors:
Stefania Lenarduzzi*, **Beatrice Spedicati***, Beatrice Alessandrini, Paola Tesolin, Alessia Paldino, Marta Gigli, Gianfranco Sinagra, Paolo Gasparini, Matteo Dal Ferro, Giorgia Girotto
*S.L. and B.S. are joined first authors

https://doi.org/10.1002/mgg3.2143

2023
Odontostomatological traits in North-Eastern Italy's isolated populations: an epidemiological cross-sectional study

Authors:
Valentina Luppieri, Alessandro Pecori, **Beatrice Spedicati***, Riccardo Schito, Lucia Pozzan, Aurora Santin, Giorgia Girotto, Milena Cadenaro, Maria Pina Concas
*B.S.: corresponding author

https://doi.org/10.3390/jcm12072746

2023
Regulator of G-Protein Signalling 9: a new candidate gene for sweet food liking?

Authors:
Catherine Anna-Marie Graham, **Beatrice Spedicati**, Giulia Pelliccione, Paolo Gasparini, Maria Pina Concas

https://doi.org/10.3390/foods12091739

2023
Identifying missing pieces in Color Vision Defects: A Genome-Wide Association Study in Silk Road populations

Authors:
Giuseppe Giovanni Nardone*, **Beatrice Spedicati***, Maria Pina Concas, Aurora Santin, Lorenzo Mazzetto, Maurizio Battaglia-Parodi, Giorgia Girotto
* G.G.N. and B.S. are joined first authors

https://doi.org/10.3389/fgene.2023.1161696

2023
Puzzling Out the Genetic Architecture of Endometriosis: Whole-Exome Sequencing and Novel Candidate Gene Identification in a Deeply Clinically Characterised Cohort

Authors:
Aurora Santin, **Beatrice Spedicati***, Anna Morgan, Stefania Lenarduzzi, Paola Tesolin, Giuseppe Giovanni Nardone, Daniela Mazzà, Giovanni Di Lorenzo, Federico Romano, Francesca Buonomo, Alessandro Mangogna, Maria Pina Concas, Gabriella Zito, Giuseppe Ricci, Giorgia Girotto
* B.S: corresponding author

https://doi.org/10.3390/biomedicines11082122

2023
Beckwith-Wiedemann syndrome and twinning: case report and brief review of literature

Authors:
Pierandrea Elefante, **Beatrice Spedicati**, Flavio Faletra, Laura Pignata, Flavia Cerrato, Andrea Riccio, Egidio Barbi, Luigi Memo, Laura Travan

doi: 10.1186/s13052-023-01530-8

2023
Gene-educational attainment interactions in a multi-population genome-wide meta-analysis identify novel lipid loci

Authors:
Lisa de las Fuentes, Karen L. Schwander, Michael R. Brown, et al. (including **Beatrice Spedicati**)

https://doi.org/10.3389/fgene.2023.1235337

2023
Which Came First? When Usher Syndrome Type 1 Couples with Neuropsychiatric Disorders

Authors:
Paola Tesolin, Aurora Santin, Anna Morgan, Stefania Lenarduzzi, Elisa Rubinato, Giorgia Girotto, **Beatrice Spedicati**

doi.org/10.3390/audiolres13060086

2024
Multi-trait analysis characterizes the genetics of thyroid function and identifies causal associations with clinical implications

Authors:
Rosalie B. T. M. Sterenborg, Inga Steinbrenner, Yong Li, et al. (including **Beatrice Spedicati**)

https://doi.org/10.1038/s41467-024-44701-9

2024
Clenching the Strings of Bruxism Etiopathogenesis: Association Analyses on Genetics and Environmental Risk Factors in a Deeply Characterized Italian Cohort

Authors:
Alessandro Pecori, Valentina Luppieri, Aurora Santin, **Beatrice Spedicati**, Stefania Zampieri, Milena Cadenaro, Giorgia Girotto, Maria Pina Concas

https://doi.org/10.3390/biomedicines12020304

2024
The Bittersweet Symphony of COVID-19: Associations between TAS1Rs and TAS2R38 Genetic Variations and COVID-19 Symptoms

Authors:
Aurora Santin*, **Beatrice Spedicati***, Alessandro Pecori, Giuseppe Giovanni Nardone, Maria Pina Concas, Gioia Piatti, Anna Menini, Giancarlo Tirelli, Paolo Boscolo-Rizzo, Giorgia Girotto
*A.S. and B.S. are joined first authors

https://doi.org/10.3390/life14020219

2024
Genome-wide analysis in over 1 million individuals of European ancestry yields improved polygenic risk scores for blood pressure traits

Authors:
Jacob M. Keaton, Zoha Kamali, Tian Xie et al. (including **Beatrice Spedicati**)

https://doi.org/10.1038/s41588-024-01714-w

2024
Proangiogenic properties of complement protein C1q can contribute to endometriosis

Authors:
Chiara Agostinis, Miriam Toffoli, Gabriella Zito, Andrea Balduit, Silvia Pegoraro, Mariagiulia Spazzapan, Lorella Pascolo, Federico Romano, Giovanni Di Lorenzo, Alessandro Mangogna, Aurora Santin, **Beatrice Spedicati**, Erica Valencic, Giorgia Girotto, Giuseppe Ricci, Uday Kishore, Roberta Bulla

https://doi.org/10.3389/fimmu.2024.1405597

2024
Understanding the genetic complexity of puberty timing across the allele frequency spectrum

Authors:
Katherine A. Kentistou, Lena R. Kaisinger, Stasa Stankovic, et al. (including **Beatrice Spedicati**)

https://doi.org/10.1038/s41588-024-01798-4

2024
A genome-wide association meta-analysis of all-cause and vascular dementia

Authors:
TheMega Vascular Cognitive Impairment and Dementia (MEGAVCID) consortium (including **Beatrice Spedicati**)

https://doi.org/10.1002/alz.14115

2024
Genome-Wide Interaction Analyses of Serum Calcium on Ventricular Repolarization Time in 125 393 Participants

Authors:
William J. Young, Peter J. van der Most, Traci M. Bartz, et al (including **Beatrice Spedicati**)

https://doi.org/10.1161/JAHA.123.034760

2024
Does It Run in Your Family? Inherited Truncating PSMD12 Variants Broaden the Phenotypic Spectrum of Stankiewicz-Isidor Syndrome

Authors:
Agnese Feresin, **Beatrice Spedicati**, Stefania Zampieri, Anna Morgan, Andrea Magnolato, Alessandra Tesser, Alberto Tommasini, Maria Teresa Bonati, Giorgia Girotto, Flavio Faletra
* B.S: corresponding author

https://doi.org/10.1002/ajmg.a.63953

2025
Expanding the Molecular Spectrum of MMP21 Missense Variants: Clinical Insights and Literature Review

Authors:
Domizia Pasquetti, Paola Tesolin, Federica Perino, Stefania Zampieri, Marco Bobbo, Thomas Caiffa, **Beatrice Spedicati**, Giorgia Girotto
* B.S.: corresponding author

https://doi.org/10.3390/genes16010062

2025
Scent of COVID-19: Whole-Genome Sequencing Analysis Reveals the Role of ACE2, IFI44, and NDUFAF4 in Long-Lasting Olfactory Dysfunction

Authors:
Beatrice Spedicati, Alessandro Pecori, Maria Pina Concas, Aurora Santin, Romina Ruberto, Giuseppe Giovanni Nardone, Andrea D'Alessandro, Giancarlo Tirelli, Paolo Boscolo-Rizzo, Giorgia Girotto

https://doi.org/10.3390/life15010056

2025
A possible association between low MBL/lectin pathway functionality and microbiota dysbiosis in endometriosis patients

Authors:
Miriam Toffoli, Giuseppina Campisciano, Aurora Santin, Silvia Pegoraro, Gabriella Zito, **Beatrice Spedicati**, Andrea Balduit, Federico Romano, Giovanni Di Lorenzo, Alessandro Mangogna, Paola Tesolin, Giuseppe Giovanni Nardone, Nunzia Zanotta, Serena Sanna, Francesca Crobu, Uday Kishore, Giuseppe Ricci, Roberta Bulla, Giorgia Girotto, Chiara Agostinis

https://doi.org/10.1016/j.lfs.2025.123427

2025
A Large-Scale Genome-wide Association Study of Blood Pressure Accounting for Gene-Depressive Symptomatology Interactions in 564,680 Individuals from Diverse Populations

Authors:
Songmi Lee, Clint L Miller, Amy R Bentley, et al. (including **Beatrice Spedicati**)

https://doi.org/10.21203/rs.3.rs-6025759/v1

2025
Prediction and prognostic role of left ventricular systolic dysfunction in family screening for dilated cardiomyopathy and non-dilated left ventricular cardiomyopathy

Authors:

Eva Del Mestre, Alessia Paldino, Carola Pio Loco Detto Gava, Ilaria Gandin, Marta Gigli, Davide Stolfo, Martina Setti, Giovanni Maria Severini, **Beatrice Spedicati**, Stefania Lenarduzzi, Giorgia Girotto, Alessandro Folgheraiter, Jacopo Giulio Rizzi, Renata Korcova, Luisa Mestroni, Marco Merlo, Matteo Dal Ferro

https://doi.org/10.1002/ejhf.3657

2025

Uncovering a Novel Pathogenic Mechanism of BCS1L in Mitochondrial Disorders: Insights from Functional Studies on the c. 38A>G Variant

Authors:
Valeria Capaci, Luisa Zupin, Martina Magistrati, Maria Teresa Bonati, Fulvio Celsi, Irene Marrone, Francesco Baldo, Blendi Ura, **Beatrice Spedicati**, Anna Morgan, Irene Bruno, Massimo Zeviani, Cristina Dallabona, Giorgia Girotto, Andrea Magnolato

https://doi.org/10.3390/ijms26083670

2025

Normal hearing function genetics: have you heard all about it? An integrated approach of genome-wide association studies and transcriptome-wide association studies in three Italian cohorts

Authors:
Aurora Santin, Giulia Pianigiani, Alessandro Gialluisi, Alessandro Pecori, **Beatrice Spedicati**, Simona Costanzo, Mariarosaria Persichillo, Francesca Bracone, Giuseppe Giovanni Nardone, Paola Tesolin, Stefania Lenarduzzi, Anna Morgan, Amalia De Curtis, Wouter van der Valk, Francis Rousset, Marta Roccio, Heiko Locher, Licia Iacoviello, Maria Pina Concas, Giorgia Girotto

https://doi.org/10.3389/fgene.2025.1522338

2025

Genetic and Phenotypic Characterization of Nexilin (NEXN)-Related Cardiomyopathy: Results From a Multicentric Study

Authors:
Maria Perotto, Alessia Paldino, Francesco Mazzarotto, Giulia Barbati, Sophie L V M Stroeks, Job A J Verdonschot, Mohammed Akhtar, Perry Elliott, Juan Pablo Ochoa, Pablo Garcia-Pavia, Fernando de Frutos, Robert Sepp, Lidia Hategan, Sanjay Prasad, Momina Yazdani, Deborah Morris-Rosendahl, Eszter Dalma Palinkas, Francesca Girolami, Iacopo Olivotto, Victoria N Parikh, Diane Fatkin, Neal Lakdawala, William J McKenna, Davide Stolfo, Marta Gigli, Francesca Brun, Chiara Collesi, Mauro Giacca, Serena Zacchigna, Giovanni Maria Severini, Stefania Lenarduzzi, **Beatrice Spedicati**, Aurora Santin, Giorgia Girotto, Paolo Gasparini, Matthew R G Taylor, Luisa Mestroni, Marco Merlo, Gianfranco Sinagra, Matteo Dal Ferro

https://doi.org/10.1016/j.jchf.2025.102529

2025

Polygenic prediction of body mass index and obesity through the life course and across ancestries

Authors:
Roelof A J Smit, Kaitlin H Wade, Qin Hui, et al. (including **Beatrice Spedicati**)

https://doi.org/10.1038/s41591-025-03827-z

2025

The homoplasmic MT-TK m.8357T > C mtDNA variant as a cause of multiorgan mitochondrial disease

Authors:
Luisa Zupin, Valeria Capaci, Maria Teresa Bonati, Eleonora Lamantea, Muhammad Suleman, Andrea Marsala, Fulvio Celsi, **Beatrice Spedicati**, Sergio Crovella, Giulia Gortani, Giorgia Girotto, Irene Bruno, Massimo Zeviani

https://doi.org/10.1016/j.mito.2025.102080

2025

Hidden in the Genome: The First Italian Family with North Carolina Macular Dystrophy Carrying a Novel PRDM13 and CCNC Duplication

Authors:
Beatrice Spedicati, Domizia Pasquetti, Aurora Santin, Stefania Zampieri, Anna Morgan, Stefania Lenarduzzi, Giuseppe Giovanni Nardone, Elisa Paccagnella, Stefania Cappellani, Laura Diplotti, Stefano Pensiero, Fulvio Parentin, Paolo Gasparini, Maurizio Battaglia Parodi, Giorgia Girotto

https://doi.org/10.3390/biomedicines13081904

2025
A Hitchhiker Guide to Structural Variant Calling: A Comprehensive Benchmark Through Different Sequencing Technologies

Authors:
Giuseppe Giovanni Nardone, Valentina Andrioletti, Aurora Santin, Anna Morgan, **Beatrice Spedicati**, Maria Pina Concas, Paolo Gasparini, Giorgia Girotto, Ivan Limongelli

https://doi.org/10.3390/biomedicines13081949

2025
Unraveling the Functional Impact of Splicing Variants in Inherited Hearing Disorders Through Minigene Splicing Assays

Authors:
Lara Emily Rosso, Giulia Pianigiani, Anna Morgan, Elisa Rubinato, Elisa Paccagnella, Stefania Lenarduzzi, Anita Wischmeijer, **Beatrice Spedicati**, Giorgia Girotto

https://doi.org/10.3390/biomedicines13092245

2025
Unveiling the spectrum of sudden cardiac death: a multidisciplinary analysis from the Friuli Venezia Giulia registry

Authors:
Davide Radaelli, Carlo Moreschi, Rossana Bussani, Maria Basciu, Carla Di Loreto, Monica Concato, Riccardo Addobbati, Martina Franzin, Maria Assunta Cova, Lorenzo Pagnan, Giorgia Girotto, Stefania Lenarduzzi, **Beatrice Spedicati**, Matteo Dal Ferro, Gianfranco Sinagra, Stefano D'Errico

https://doi.org/10.3389/fcvm.2025.1651235

ABSTRACTS SUBMITTED TO NATIONAL AND INTERNATIONAL CONFERENCES AND MEETINGS

2019
The relevant role of Italian genetic isolates for the study of Human Knockouts

Abstract submitted to the European Human Genetics Conference 2019
Authors:
F. Sirchia, M. Cocca, F. Faletra, G. Girotto, **B. Spedicati**, A. Morgan, R. Palmisano, C. Barbieri, D. Toniolo, P. Gasparini

2020
Impairments of smell perception: the role of sex, age and genes

Abstract submitted to the European Human Genetics Conference 2020
Authors:
M.P. Concas, T. Battistuzzi, M. Cocca, **B. Spedicati**, G. Pelliccione, P. Gasparini, G. Girotto

2020
The role of bi-allelic Loss of Function variants in Olfactory Receptor genes on the perception of smell

Abstract submitted to the XXIII SIGU National Congress
Authors:
M.P. Concas, M. Francescatto, T. Battistuzzi, **B. Spedicati**, G. Pelliccione, A. Morgan, A. Feresin, M. Cocca, P. Gasparini, G. Girotto

2020
The role of POGZ in neurodevelopmental disorders: from molecular diagnosis to a possible therapeutic approach

Abstract submitted to the XXIII SIGU National Congress
Authors:
G. Girotto, **B. Spedicati**, C. Romano, L. Garavelli, M. Dentici, N. Specchio, P. Alfieri, P. Grammatico, G. Trimarchi, M. Tartaglia, P. Gasparini, F. Faletra, A. Feresin

2020
Multiplex Ligation Probe Amplification (MLPA) and Whole Exome Sequencing (WES) revealed new alleles/genes in a subset of 214 Italian families affected by Non-Syndromic Hearing Loss (NSHL)

Abstract submitted to the XXIII SIGU National Congress
Authors:
A. Morgan, F. Faletra, S. Lenarduzzi, M. La Bianca, G. Pelliccione, **B. Spedicati**, A. Feresin, D. Mazzà, A. Sensi, C. Graziano, M. Seri, U. Ambrosetti, P. Gasparini, G. Girotto.

2021
From molecular diagnosis to a possible therapeutic approach: a proposal for POGZ-related Neurodevelopmental Disorder

Abstract submitted to the Genomics of Rare Disease Virtual conference 2021
Authors:
A. Feresin, **B. Spedicati**, C. Romano, L. Garavelli, M. Dentici, N. Specchio, P. Alfieri, P. Grammatico, G. Trimarchi, M. Tartaglia, P. Gasparini, G. Girotto, F. Faletra, M. Bonati

2021
Multiplex Ligation Probe Amplification (MLPA) and Whole Exome Sequencing (WES) for the molecular diagnosis of Non-Syndromic Hearing Loss (NSHL): the results of a cohort of 214 Italian families.

Abstract submitted to the Association for Research in Otolaryngology - Midwinter meeting 2021
Authors:
A. Morgan, F. Faletra, S. Lenarduzzi, M. La Bianca, G. Pelliccione, **B. Spedicati**, A. Feresin, D. Mazzà, A. Sensi, C. Graziano, M. Seri, U. Ambrosetti, P. Gasparini, G. Girotto.

2021
Genetic dissection of Cloninger's Temperament and Character Inventory, TCI, in an Italian isolate

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Abstract submitted to the European Human Genetics Conference 2021
Authors:
M. Concas, A. Minelli, S. Aere, F. Serra, A. Morgan, **B. Spedicati**, G. Morgante, M. Cocca, M. Gennarelli, P. Gasparini, G. Girotto

2021
The portray of the Italian cohort of patients with variants in POGZ: new care opportunities from a deep genotyping and phenotyping

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Abstract submitted to the European Human Genetics Conference 2021
Authors:
A. Feresin, **B. Spedicati**, G. Pelliccione, C. Romano, L. Garavelli, M. Dentici, N. Specchio, P. Alfieri, P. Grammatico, G. Trimarchi, M. Baldassarri, A. Renieri, R. Milone, F. Faletra, G. Cossu, G. Girotto, M. Tartaglia, P. Gasparini, M.T. Bonati.

2021
GJB2 sequencing, Multiplex Ligation Probe Amplification (MLPA) and Whole Exome Sequencing (WES) for the molecular diagnosis of Non-Syndromic Hearing Loss (NSHL): the experience of a cohort of 277 Italian families.

Abstract submitted to the European Human Genetics Conference 2021
Authors:
A. Morgan, F. Faletra, S. Lenarduzzi, M. La Bianca, G. Pelliccione, **B. Spedicati**, A. Feresin, D. Mazzà, A. Alberto, C. Graziano, M. Seri, U. Ambrosetti, P. Gasparini, G. Girotto

2021
Human knockouts of olfactory receptors genes and smell perception impairment in a large Italian cohort

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Abstract submitted to the European Human Genetics Conference 2021
Authors:
P. Tesolin, M. Concas, M. Cocca, M. Francescatto, A. Luglio, **B. Spedicati**, A. Feresin, A. Morgan, P. Gasparini, G. Girotto

2021
An X-linked dominant condition with incomplete penetrance? A familiar case of SOX3 duplication within three generations of healthy subjects and affected fetuses

---

Abstract submitted to the XXIV SIGU National Congress
Authors:
A. Feresin, S. Cappellani, S. Ulivi, **B. Spedicati**, G. Morgante, A. Luglio, E. Rubinato, M.T. Bonati, F. Faletra, G. Girotto, T. Stampalija, D. Adamo Pio, P. Gasparini

2021
Whole Exome Sequencing for fetuses with structural anomalies: which contribute in the postmortem diagnostic pathway?

Abstract submitted to the XXIV SIGU National Congress
Authors:
A. Feresin, **B. Spedicati**, G. Morgante, A. Luglio, S. Sarah, M. La Bianca, G. Pelliccione, I. Fantasia, T. Stampalija, F.M. Murru, A. Morgan, E. Rubinato, M.T. Bonati, F. Faletra, G. Girotto, R. Bussani, P. Gasparini

2021
When short stature is not a small thing: a medical history that began with GH deficiency hypostaturism

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Abstract submitted to the XXIV SIGU National Congress
Authors:
A. Luglio, M.T. Bonati, D. Di Bella, S. Magri, E. Sarto, L. Nanetti, F. Faletra, A. Feresin, A. Morgan, G. Morgante, L. Musante, E. Rubinato, **B. Spedicati**, G. Girotto, F. Taroni, P. Gasparini

2021
A novel PRKAR1B variant as a cause of intellectual disability and hyperphagia

Abstract submitted to the XXIV SIGU National Congress
Authors:
G. Morgante, M.T. Bonati, F. Faletra, A. Feresin, A. Luglio, A. Morgan, L. Musante, E. Rubinato, **B. Spedicati**, G. Girotto, P. Gasparini

2022
PSMD12 haploinsufficiency is not simply a Neurodevelopmental Disorder

Abstract submitted to the European Human Genetics Conference 2022
Authors:
A. Feresin, M.T. Bonati, F. Faletra, G. Girotto, A. Luglio, A. Morgan, G. Morgante, E. Rubinato, **B. Spedicati**, A. Tesser, A. Tommasini, P. Gasparini

2022
PERCHING syndrome: extremely rare and complicate clinical diagnnosis

Abstract submitted to the European Human Genetics Conference 2022
Authors:
A. Luglio, E. Rubinato, M.T. Bonati, F. Faletra, A. Feresin, A. Morgan, G. Morgante, L. Musante, **B. Spedicati**, G. Girotto, P. Gasparini

2022
Accurate clinical evaluation and high throughput technologies for the molecular characterization of hereditary hearing loss in a large cohort of Italian patients

Abstract submitted to the European Human Genetics Conference 2022
Authors:
A. Morgan, S. Lenarduzzi, **B. Spedicati**, P. Tesolin, A. Santin, E. Rubinato, G. Girotto

2022
Whole Exome Sequencing (WES) in unravelling complex cases of bicuspid aortic valve

Abstract submitted to the European Human Genetics Conference 2022
Authors:
G. Morgante, M. T. Bonati, F. Faletra, A. Morgan, E. Rubinato, A. Feresin, A. Luglio, **B. Spedicati**, G. Girotto, P. Gasparini

2022
Shedding light on Endometriosis (EM) disease: Whole Exome Sequencing (WES) and new genes discovery in a fully clinical characterized Italian cohort

Abstract submitted to the European Human Genetics Conference 2022
Authors:
A. Santin, P. Tesolin, **B. Spedicati**, G. Zito, F. Romano, A. Morgan, M. Concas, G. Ricci, G. Girotto

2022
The sense of the genetic diversity: a comparison between medicine and arts

Abstract submitted to the XXV SIGU National Congress
Authors:
B. Alessandrini, **B. Spedicati**, A. Luglio, M. Della Monica, G. Zampino, G. Scarano, P. Gasparini, G. Girotto, L. Memo

2022
Whole Exome Sequencing in unravelling complex cases of bicuspid aortic valve

Abstract submitted to the XXV SIGU National Congress
Authors:
F. Faletra, M.T. Bonati, A. Morgan, E. Rubinato, A. Feresin, A. Luglio, **B. Spedicati**, G. Girotto, P. Gasparini

2022
Not only a Klippel-Feil, nor a tube defect: a novel Italian family with V239I VANGL1

Abstract submitted to the XXV SIGU National Congress
Authors:
A. Feresin, A. Luglio, **B. Spedicati**, A. Morgan. M.T. Bonati, F. Faletra, G. Girotto, P. Gasparini, E. Rubinato

2022
Whole Exome Sequencing for fetal anomalies: where are we and where are we going? A single center experience and a review of the current practices

Abstract submitted to the XXV SIGU National Congress
Authors:

A. Feresin, A. Luglio, **B. Spedicati**, A. Morgan, M.T. Bonati, F. Faletra, E. Rubinato, T. Stampalija, R. Bussani, F. Murru, G. Girotto, P. Gasparini

2022

Deep phenotyping and high throughput sequencing technologies for the molecular diagnosis of hereditary hearing loss in an Italian cohort of patients

Abstract submitted to the XXV SIGU National Congress
Authors:
A. Morgan, S. Lenarduzzi, **B. Spedicati**, G.G. Nardone, P. Tesolin, A. Santin, E. Rubinato, G. Girotto

2022

Orthodontic measurements in isolated populations from North - Eastern Italy: an epidemiological and genetics study

Abstract submitted to the XXV SIGU National Congress
Authors:
A. Pecori, V. Luppieri, **B. Spedicati**, R. Schito, M. Cadenaro, G. Girotto, M. P. Concas

2022

The challenge of non-syndromic mimics: our experience with hereditary hearing loss

Abstract submitted to the XXV SIGU National Congress
Authors:
E. Rubinato, A. Morgan, F. Faletra, A. Feresin, **B. Spedicati**, G. Girotto

2022

Normal Hearing Function (NHF) and Age-Related Hearing Loss (ARHL): new candidate genes identification from Genome-Wide Association Studies (GWAS) in Moli-sani cohort

Abstract submitted to the XXV SIGU National Congress
Authors:
A. Santin, S. Lenarduzzi, G.G. Nardone, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Giallusi, L.Iacoviello, P. Gasparini, M.P. Concas, G. Girotto

2022

Novel genetic insights into Endometriosis (EM) disease: the first Italian Whole-Exome Sequencing (WES) study

Abstract submitted to the XXV SIGU National Congress
Authors:
A. Santin, P. Tesolin, **B. Spedicati**, G.G. Nardone, G. Zito, F. Romano, G. Di Lorenzo, A. Morgan, M.P. Concas, G. Ricci, G. Girotto

2022

Non-syndromic or syndromic hearing loss? our experience with the challenge of non-syndromic mimics

Abstract submitted to the Symposium & 57 Inner Ear Biology Workshop – IEB 2022
Authors:
E. Rubinato, A. Morgan, F. Faletra, A. Feresin, **B. Spedicati**, G. Girotto

2022

Deepening the genetics of hearing: Genome-Wide Association Studies (GWAS) on Moli-sani cohort

Abstract submitted to the Symposium & 57 Inner Ear Biology Workshop – IEB 2022
Authors:
A. Santin, S. Lenarduzzi, G. G. Nardone, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, L. Iacoviello, P. Gasparini, M. P. Concas, G. Girotto

2022

Genetic diseases in the arts

Abstract submitted to the 9th Congress of the Genetic society of Slovenia - Proceedings of Genetika 2022
Authors:
B. Alessandrini, **B. Spedicati**, A. Luglio, M. Della Monica, G. Zampino, G. Scarano, G. Girotto, L. memo, P. Gasparini

2023

Genetic syndromes hiding behind art masterpieces: an intriguing journey between medicine and arts

Abstract submitted to the European Human Genetics Conference 2023
Authors:
G. Girotto, **B. Spedicati**, B. Alessandrini, M. Della Monica, G. Zampino, G. Scarano, P. Gasparini, L. Memo

2023

Whole Genome Sequencing (WGS) for the molecular diagnosis of Hereditary Hearing Loss (HHL): the underestimated role of Structural Variants (SVs)

Abstract submitted to the European Human Genetics Conference 2023
Authors:
G. G. Nardone, A. Santin, A. Morgan, **B. Spedicati**, M. P. Concas, G. Girotto

2023

Genetic factors involved in bruxism: the first Genome-Wide Association Study (GWAS) in isolated populations from North-Eastern Italy

Abstract submitted to the European Human Genetics Conference 2023
Authors:
A. Pecori, V. Luppieri, **B. Spedicati**, A. Santin, R. Schito, M. Cadenaro, G. Girotto, M. P. Concas

2023

Puzzling out the genetic landscape of Hearing Function (HF): a combined approach of Genome-Wide Association Studies (GWAS) and Transcriptome-Wide Association Studies (TWAS)

Abstract submitted to the European Human Genetics Conference 2023
Authors:
A. Santin, S. Lenarduzzi, G. G. Nardone, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, L. Iacoviello, P. Gasparini, M. P. Concas, G. Girotto

2023

Rapid whole exome sequencing (rWES) in neonatal care in an Italian maternnal-children hospital

Abstract submitted to the European Human Genetics Conference 2023
Authors:
C. Sbaffi, **B. Spedicati**, E. Rubinato, F. Faletra, G. Pelliccione, S. Zampieri, A. Morgan, G. Girotto

2023

Genetic variants in RGS9 are associated with sweet food liking and eating behaviour in Italian cohorts

Abstract submitted to Pangborn Sensory Science Symposium 2023
Authors:
C. A-M. Graham, **B. Spedicati**, G. Pelliccione, P. Gasparini, M. P. Concas

2023

iPSC-derived Inner Ear Organoids and CRISPR/Cas9-mediated Genome-Editing: a Disease Model System to Characterize a Novel MYO5C Mutation as Putative Cause of Hearing Loss

Abstract submitted to the Symposium & 58 Inner Ear Biology Workshop – IEB 2023
Authors:
G. Pianigiani, A. Morgan, A. Abidi, S. Alonso Jimenez, M. Generali, **B. Spedicati**, P. Gasparini, G. Girotto, M. Roccio

2023

Hearing Function (HF): a Combined Approach of Genome-Wide Association Studies (GWAS) Meta-analysis, Gene Expression and Transcriptome-Wide Association Studies (TWAS) Analysis

Abstract submitted to the Symposium & 58 Inner Ear Biology Workshop – IEB 2023
Authors:
A. Santin, S. Lenarduzzi, G.G. Nardone, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, P. Gasparini, M.P. Concas, G. Girotto

2023

The impact of Whole Exome Sequencing for the molecular diagnosis of fetuses with malformations: the experience of a referral Italian hospital

Abstract submitted to the XXVI SIGU National Congress
Authors:
A. Feresin, S. Zampieri, A. Morgan, M.T. Bonati, E. Paccagnella, E. Rubinato, D. Mazzà, **B. Spedicati**, T. Stampalija, M. Pinamonti, R. Bussani, P. Gasparini, F. Murru, G. Girotto

2023

Incidentalomi in geni associati a rischio oncologico in pazienti candidati ad esoma per malattie cardiologiche

Abstract submitted to the XXVI SIGU National Congress
Authors:
D. Mazzà, S. Lenarduzzi, A. Morgan, S. Zampieri, **B. Spedicati**, E. Paccagnella, A. Feresin, M. T. Bonati, E. Rubinato, M. Dal Ferro, A. Paldino, S. D'Errico, G. Sinagra, P. Gasparini, G. Girotto

2023

Unravelling the genetic bases of Hearing Loss (HL): deep phenotyping and high throughput sequencing technologies in an Italian Cohort of patients

Abstract submitted to the XXVI SIGU National Congress
Authors:
A. Morgan, **B. Spedicati**, A. Santin, G. G. Nardone, S. Lenarduzzi, E. Paccagnella, E. Rubinato, G. Girotto

2023

Structural Variation in short-read Whole Genome Sequencing (srWGS): towards a more accurate detection for clinical practice implementation

Abstract submitted to the XXVI SIGU National Congress
Authors:
G. G. Nardone, V. Andrioletti, A. Santin, A. Morgan, **B. Spedicati**, M. P. Concas, I. Limongelli, G. Girotto

2023

Whole Genome Sequencing (WGS) in a highly characterized Italian cohort: assessing the role of Loss of Function variants (LoF) in COVID-19 persistent chemosensory dysfunction

Abstract submitted to the XXVI SIGU National Congress
Authors:
G. G. Nardone, **B. Spedicati**, A. Pecori, A. Santin, A. Morgan, M. P. Concas, P. Gasparini, P. Boscolo-Rizzo, G. Girotto

2023

Expanding the molecular and clinical phenotype of the PIG-family disorders

Abstract submitted to the XXVI SIGU National Congress
Authors:
E. Paccagnella, A. Feresin, M. T. Bonati, A. Morgan, S. Zampieri, **B. Spedicati**, I. Bruno, M. Carrozzi, C. Zanus, G. Girotto, L. Musante

2023

Exploring new genes and risk factors factors associated with bruxism in Italian samples

Abstract submitted to the XXVI SIGU National Congress
Authors:
A. Pecori, S. Zampieri, V. Luppieri, **B. Spedicati**, A. Santin, M. Cadenaro, G. Girotto, M. P. Concas

2023

Endometriosis (EM): still an unsolved genetic dilemma? Whole-Exome Sequencing (WES) analysis and gene discovery in a highly characterized cohort

Abstract submitted to the XXVI SIGU National Congress
Authors:
A. Santin, **B. Spedicati**, A. Morgan, S. Lenarduzzi, P. Tesolin, G. G. Nardone, S. Camarda, H. Stevens, D. Mazzà, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, M. P. Concas, G. Zito, G. Ricci, G. Girotto

2023

Hearing Function (HF) genetics: you have heard only a little about. Genome-Wide Association Studies (GWAS) meta-analyses and Transcriptome-Wide Association Studies (TWAS) analyses on Italian cohorts

Abstract submitted to the XXVI SIGU National Congress
Authors:
A. Santin, S. Lenarduzzi, G.G. Nardone, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, L. Iacoviello, P. Gasparini, M.P. Concas, G. Girotto

2023

The new era of arts and genetics: a microscopic study of macroscopic representations

Abstract submitted to the XXVI SIGU National Congress
Authors:
P. Tesolin, **B. Spedicati**, A. Feresin, M. Della Monica, G. Zampino, G. Scarano, P. Gasparini, L. Memo, G. Girotto

2023

Integrated approach for the molecular characterization of 440 Italian patients affected by different forms of hereditary cardiovascular diseases

Abstract submitted to the XXVI SIGU National Congress
Authors:
S. Zampieri, S. Lenarduzzi, A. Paldino, **B. Spedicati**, D. Mazzà, M. Dal Ferro, G. Sinagra, P. Gasparini, G. Girotto

2023

Whole Genome Sequencing (WGS) in COVID-19 persistent chemosensory dysfunction: assessing the role of Loss of Function variants (LoFs) in a highly characterized Italian cohort

Abstract submitted to the American Society of Human Genetics Annual Meeting 2023
Authors:
G. G. Nardone, **B. Spedicati**, A. Pecori, A. Santin, A. Morgan, M. P. Concas, G. Tirelli, P. Gasparini, P. Boscolo-Rizzo, G. Girotto

2023

Unraveling the genetic enigma of Endometriosis: novel insights and gene discovery on a deeply characterized cohort

Abstract submitted to the American Society of Human Genetics Annual Meeting 2023
Authors:
A. Santin, **B. Spedicati**, A. Morgan, S. Lenarduzzi, P. Tesolin, G. G. Nardone, D. Mazzà, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, M. P. Concas, G. Zito, G. Ricci, G. Girotto

2024

The value of SNP - arrays analysis in pregnancy losses: a retrospective study

Abstract submitted to the Human Genome Meeting 2024
Authors:
S. Cappellani, A. P. d'Adamo, **B. Spedicati**, E. Paccagnella, S. Zampieri, D. Favretto, G. Turchetto, D. De Santo, G. Maso, S. Ulivi, G. Girotto, P. Gasparini

2024

Integrated approach for the molecular characterization of 486 Italian patients affected by different forms of hereditary cardiovascular diseases

Abstract submitted to the Human Genome Meeting 2024
Authors:
G. Girotto, S. Zampieri, A. Paldino, **B. Spedicati**, D. Daniela, G. Turchetto, M. Dal Ferro, G. Sinagra, S. Lenarduzzi

2024

Optimizing Structural Variant Calling: towards a robust and reliable detection from Whole Genome Sequencing (WGS)

Abstract submitted to the Human Genome Meeting 2024
Authors:
G. G. Nardone, V. Andrioletti, A. Santin, A. Morgan, **B. Spedicati**, M. P. Concas, I. Limongelli, G. Girotto

2024

Investigation of environmental and genetic factors associated with bruxism in five genetically isolated populations from North-Eastern Italy

Abstract submitted to the Human Genome Meeting 2024
Authors:
A. Pecori, V. Luppieri, A. Santin, **B. Spedicati**, C. Bon ,S. Zampieri ,M. Cadenaro, G. Girotto, M. P. Concas

2024

Connecting the dots of endometriosis disease: the role of genetics, diet, and microbiome

Abstract submitted to the Human Genome Meeting 2024
Authors:
A. Santin, A. Pecori, **B. Spedicati**, A. Morgan, S. Lenarduzzi, P. Tesolin, G. G. Nardone, S. Camarda, H. Stevens, G. Pianigiani, L. E. Rosso, E. Vinerbi, C. Bon, D. Mazzà, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, G. Campisciano, G. Zito, S. Sanna, M. P. Concas, G. Ricci, G. Girotto

2024

The impact of Whole Exome Sequencing for the molecular diagnosis of fetal malformations: the experience of a referral Italian hospital

Abstract submitted to the Human Genome Meeting 2024
Authors:
S. Zampieri, A. Feresin, A. Morgan, E. Paccagnella, E. Rubinato, D. Mazzà, **B. Spedicati**, T. Stampalija, M. Pinamonti, R. Bussani, P. Gasparini, F. Murru, G. Girotto

2024

Molecular basis of hereditary cardiovascular diseases (hCVDs) in a cohort of 486 Italian patients: a multi-step approach

Abstract submitted to the European Human Genetics Conference 2024
Authors:
G. Girotto, S. Zampieri, A. Paldino, **B. Spedicati**, G. Turchetto, D. Mazzà, M. Dal Ferro,G. Sinagra, S. Lenarduzzi

2024
Modeling genetic forms of hearing loss with inner ear organoids

Abstract submitted to the European Human Genetics Conference 2024
Authors:
G. Pianigiani, A. Morgan, M. Troian, G. Turchetto, **B. Spedicati**, M. Roccio, G. Girotto

2024
The molecular riddle of Endometriosis (EM) disorder: filling in the blanks between genetics, diet, and microbiome

Abstract submitted to the European Human Genetics Conference 2024
Authors:
A. Santin, **B. Spedicati**, A. Morgan, S. Lenarduzzi, P. Tesolin, A. Pecori, G. G. Nardone, S. Camarda, H. Stevens, G. Pianigiani, L. E. Rosso, C. Bon, D. Mazzà, E. Vinerbi, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, G. Campisciano, G. Zito, S. Sanna, M. P. Concas, G. Ricci, G. Girotto

2024
The implementation of Whole Exome Sequencing in the molecular diagnosis of fetal structural anomalies: the experience of a referral Italian hospital

Abstract submitted to the European Human Genetics Conference 2024
Authors:
S. Zampieri, A. Feresin, A. Morgan, E. Paccagnella, E. Rubinato, D. Mazzà, F. Perino, A. Saramin, T. Stampalija, M. Pinamonti, G. Turchetto, R. Bussani, **B. Spedicati**, F. Murru, G. Girotto

2024
The Hidden Truth of Hereditary Hearing Loss: Gaining Insight into the genetic basis of Non-syndromic Mimics

Abstract submitted to the Symposium & 57 Inner Ear Biology Workshop – IEB 2024
Authors:
P. Tesolin, **B. Spedicati**, A, Morgan, S. Lenarduzzi, E. Rubinato, A. Santin, M. Troian, D. Marangoni, G. Girotto

2024
Non-invasive prenatal testing (NIPT) for trisomies 13, 18, 21 and sex chromosome aneuploidies: outcomes and clinical experience from 447 consecutive pregnancies

Abstract submitted to the XXVII SIGU National Congress
Authors:
S. Cappellani, D. Pasquetti, S. Ulivi, A. Morgan, D. Favretto, G. Turchetto, **B. Spedicati**, A. P. D'Adamo, S. Zampieri, G. Girotto

2024
Clinical application of Nanopore sequencing in the molecular diagnosis of genetic disorders

Abstract submitted to the XXVII SIGU National Congress
Authors:
S. Lenarduzzi, A. Morgan, M. Troian, S. Zampieri, G. G. Nardone, C. Bon, **B. Spedicati**, P. Gasparini, G. Girotto

2024
Structural Variant (SV) calling optimization: towards a robust and reliable detection from short read Whole Genome Sequencing (srWGS) data

Abstract submitted to the XXVII SIGU National Congress
Authors:
G. G. Nardone, V. Andrioletti, A. Santin, A. Morgan, **B. Spedicati**, M. Troian, M. P. Concas, I. Limongelli, G. Girotto

2024
Functional Characterization of Splicing Variants in Syndromic and Non-Syndromic Hereditary Hearing Loss (HHL) Using Minigene Splicing Assays

Abstract submitted to the XXVII SIGU National Congress
Authors:
L. E. Rosso, A. Morgan, E. Rubinato, **B. Spedicati**, G. Girotto, G. Pianigiani

2024
Age-Related Hearing Loss (ARHL) genetics at a corner: a GWAS meta-analysis on Italian cohorts

Abstract submitted to the XXVII SIGU National Congress
Authors:
A. Santin, S. Lenarduzzi, G.G. Nardone, A. Pecori, **B. Spedicati**, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, L. Iacoviello, P. Gasparini, M.P. Concas, G. Girotto

2024
Inside-out Endometriosis disease: a 360°-degree view on clinics, genetics, microbiome and diet

Abstract submitted to the XXVII SIGU National Congress
Authors:
A. Santin, A. Pecori, **B. Spedicati**, A. Morgan, S. Lenarduzzi, P. Tesolin, G. G. Nardone, S. Camarda, H. Stevens, G. Pianigiani, L. E. Rosso, E. Vinerbi, C. Bon, D. Mazzà, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, G. Campisciano, G. Zito, S. Sanna, M. P. Concas, G. Ricci, G. Girotto

2024
Whole genome sequencing (WGS) in Italian genetic isolates reveals a new CYP2D6 allele

Abstract submitted to the XXVII SIGU National Congress
Authors:
P. Tesolin, G. G. Nardone, A. Pecori, M. Troian, A. Santin, L. E. Rosso, G. Pianigiani, **B. Spedicati**, G. Girotto

2024
The clinical utility of Whole Exome Sequencing (WES) in the molecular diagnosis in a cohort of 89 fetuses with structural anomalies: the experience of a referral Italian hospital

Abstract submitted to the XXVII SIGU National Congress
Authors:
S. Zampieri, A. Morgan, E. Paccagnella, E. Rubinato, A. Feresin, D. Mazzà, F. Perino, A. Saramin, T. Stampalija, M. Pinamonti, G. Turchetto, R. Bussani, F. Murru, **B. Spedicati**, G. Girotto

2024
Genetics of Hereditary Hearing Loss (HHL): a complex and hidden world

Abstract submitted to the 34th Politzer Society meeting
Authors:
G. Girotto, S. Lenarduzzi, M. Troian, G. Pianigiani, E. Rubinato, A. Morgan, **B. Spedicati**

2025
When appearances are deceptive: Whole Exome Sequencing (WES) reveals the presence of Non-Syndromic Mimics (NSMs)

Abstract submitted to the Young Research Meeting on ENT & Audiology and Hearing Neuroscience
Authors:
G Girotto, S. Lenarduzzi, A. Morgan, G. Pianigiani, E. Rubinato, D. Marangoni, **B. Spedicati**

2025
A Rare Case of Hypertrophic-Restrictive Cardiomyopathy with Extensive Myocardial and Valvular Calcifications: Whole-Exome Sequencing (WES) Analysis of an Uncommon Clinical Phenomenon

Abstract submitted to the European Human Genetics Conference 2025
Authors:
L. Bortot, **B. Spedicati**, A. Paldino, S. Lenarduzzi, A. Santin, M. Dal Ferro, G. Sinagra, G.Girotto

2025
Exploring the genetics of molar incisor hypomineralization (MIH): a molecular study on candidate genes involved in human dental enamel development

Abstract submitted to the European Human Genetics Conference 2025
Authors:
S. Frisari, S. Zampieri, V. Luppieri, M.P. Concas, **B. Spedicati**, A. Santin, A. Pecori, M. Cadenaro, G. Girotto

2025
Whole exome sequencing (WES) increases the molecular diagnosis of 106 foetuses with structural anomalies negative at chromosomal microarray analysis

Abstract submitted to the European Human Genetics Conference 2025
Authors:
V.R. Gasparini, **B. Spedicati**, G. Pianigiani, A. Santin, I. Falcomer, D. Mazzà, E. Paccagnella, D. Pasquetti, E. Rubinato, C. Granata, F.M. Murru, M. Pinamonti, R. Bussani, I. Fantasia, T. Stampalija, P. Gasparini, S. Zampieri, G. Girotto

2025
A Hitchhiker guide to Structural Variant (SVs) Calling: a comprehensive benchmark through short-reads (srWGS) and long-reads (lrWGS) sequencing technologies

Abstract submitted to the European Human Genetics Conference 2025
Authors:
G.G. Nardone, V. Andrioletti, A. Santin, A. Morgan, **B. Spedicati**, M. Troian, M.P. Concas, I. Limongelli, G. Girotto

2025
Functional Characterization of Splicing Variants in Syndromic and Non-Syndromic Hereditary Hearing Loss (HHL) Using Minigene Splicing Assays

Abstract submitted to the European Human Genetics Conference 2025
Authors:
L.E. Rosso, A. Morgan, E. Rubinato, **B. Spedicati**, G. Girotto, G. Pianigiani

2025
Endometriosis (EM) symptoms and extra-oral taste-associated genes (TGs): not only a matter of taste

Abstract submitted to the European Human Genetics Conference 2025
Authors:
A. Santin, A. Pecori, G.G. Nardone, **B. Spedicati,** S. Lenarduzzi, P. Tesolin, A. Morgan, S. Camarda, H. Stevens, L. Fasoli, D. Mazzà, G.Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, G. Zito, M. P. Concas, G. Ricci, G. Girotto

2025
Art and Genetics: we are not different, we are unique

Abstract submitted to the European Human Genetics Conference 2025
Authors:
P. Tesolin, D. Pasquetti, A. Santin, **B. Spedicati**, L. Memo, G. Girotto

2025
Whole genome sequencing (WGS): a powerful tool to gain novel insight into the CYP2D6 gene

Abstract submitted to the European Human Genetics Conference 2025
Authors:
S. Zampieri, P, Tesolin, G.G. Nardone, A. Pecori, M.P. Concas, M. Troian, A. Santin, L.E. Rosso, G. Pianigiani, **B. Spedicati**, G. Girotto

2025
Identification Of Three Novel Brain-Expressed Genes Potentially Associated With Bruxism

Abstract submitted to the 103rd General Session & Exhibition of the IADR
Authors:
V. Luppieri, A. Pecori, A. Santin, **B. Spedicati**, S. Zampieri, M.P. Concas, G. Girotto, M. Cadenaro

2025
Beyond the curtains of Non-syndromic Hereditary Hearing Loss (NSHHL): Whole Exome Sequencing (WES) reveals the presence of Non-Syndromic Mimics (NSMs)

Abstract submitted to the CORLAS Annual Meeting 2025
Authors:
G Girotto, S. Lenarduzzi, A. Morgan, G. Pianigiani, E. Rubinato, D. Marangoni, **B. Spedicati**

2025
Unlocking Uniparental Disomy (UPD): An innovative genetic approach using family trio analysis

Abstract submitted to the XXVIII SIGU National Congress
Authors:
S. Cappellani, R. Moura, S. Ulivi, S. Zampieri, D. Favretto, G. Turchetto, E. Paccagnella, E. Rubinato, D. Pasquetti, **B. Spedicati**, P. Gasparini, G. Girotto, A. P. d'Adamo

2025
A Hitchhiker guide to Structural Variant (SV) Calling: a comprehensive benchmark through short-reads (srWGS) and long-reads (lrWGS) sequencing technologies

Abstract submitted to the XXVIII SIGU National Congress
Authors:
G.G. Nardone, V. Andrioletti, A. Santin, A. Morgan, **B. Spedicati**, M.P. Concas, G. Girotto, I. Limongelli

2025
Exploring the gut-pelvis connection: how microbiota and taste receptor genes (TGs) influence endometriosis (EM) symptoms

Abstract submitted to the XXVIII SIGU National Congress
Authors:
L. E. Rosso, G.G. Nardone, A. Santin, **B. Spedicati**, P. Tesolin, A. Pecori, M. P. Concas, D. Mazzà, G. Di Lorenzo, F. Romano, F. Buonomo, A. Mangogna, G. Campisciano, G. Zito, G. Ricci, G. Girotto

2025
Exploring the Role of SNPs in Genes Related to Sweet Taste Perception and Intake (SweetG) in Primary Ciliary Dyskinesia (PCD) and Its Clinical Manifestations

Abstract submitted to the XXVIII SIGU National Congress
Authors:
R. Ruberto, G. Piatti, A. Santin, S. Camarda, G. G. Nardone, A. Pecori, P. Tesolin, E. Tassin, **B. Spedicati**, G. Girotto, M. P. Concas

2025
Hidden Variants: Decoding Endometriosis (EM) via Whole Exome Sequencing (WES)

Abstract submitted to the XXVIII SIGU National Congress
Authors:
A. Santin, L. Fasoli, M. Toffoli, S. Pegoraro, P. Tesolin, L. E. Rosso, A. Badalini, **B. Spedicati,** A. Morgan, S. Lenarduzzi, G.G. Nardone, G. Pianigiani, D. Mazzà, G. Zito, G. Di Lorenzo, F. Romano, A. Mangogna, F. Buonomo, M. P. Concas, G. Ricci, C. Agostinis, G. Girotto

2025
Unravelling the Genetic Basis of Molar-Incisor Hypomineralization (MIH): a molecular study on candidate genes involved in dental enamel formation

Abstract submitted to the XXVIII SIGU National Congress
Authors:
S. Zampieri, S. Frisari, V. Luppieri, M.P. Concas, **B. Spedicati**, A. Santin, A. Pecori, M. Cadenaro, G. Girotto

2025
Longitudinal Evaluation of the Psychosocial Impact of Genetic Testing for Hereditary Hearing Loss (HHL) using a Patient-Based Survey Approach

Abstract submitted to the American Society of Human Genetics Annual Meeting 2025
Authors:
D. Pasquetti, **B. Spedicati**, S. Zampieri, A. Morgan, G. Pianigiani, E. Rubinato, F. Perino, A. Pecori, M. P. Concas, L. Pellegrini, U. Albert, G. Girotto

PARTICIPATION TO CONFERENCES, SEMINARS AND MEETINGS

24/03/2018
American Heart Association Basic Life Support (CPR and AED) program – BLSD for healthcare providers course

Ordine dei Medici Chirurghi e Odontoiatri della provincia di Trieste

01/02/2019 – 31/12/2019
Diagnosi prenatale: processi di integrazione multiprofessionale

I.R.C.C.S. "Burlo Garofolo" - Trieste

15/05/2019
Sindromi da iperaccrescimento: pathway e clinica

I.R.C.C.S. "Burlo Garofolo" - Trieste

12/12/2019 – 13/12/2019
New frontiers in research, diagnostics and therapies

I.R.C.C.S. "Burlo Garofolo" - Trieste

02/2020
BLSD for healthcare providers course

I.R.C.C.S. "Burlo Garofolo" - Trieste

24/02/2020 – 28/02/2021
Discussione di casi clinici e diagnostici complessi in genetica medica

I.R.C.C.S. "Burlo Garofolo" - Trieste

30/03/2021
Webinar on Cardiomyopathies

I.R.C.C.S. "Burlo Garofolo" - Trieste

15/02/2022
10 Tips for Preparing a Succesfull Manuscript with an ACS Editor

University of Trieste

11/04/2022
The sixth taste

I.R.C.C.S. "Burlo Garofolo" - Trieste

22/04/2022
Power Point: presentations for communicating your research

University of Trieste

27/04/2022 – 28/04/2022
Banche dati, stili citazionali e ricerca delle fonti in ambito tecnico-scientifico e biomedico

University of Trieste

05/2022 – 06/2022
English for research purposes: exploring the features of written and spoken academic discourse

University of Trieste

20/10/2022
Discovery of White-Sutton Syndrome and Recent Advances in Research

University of Trieste - I.R.C.C.S. "Burlo Garofolo" - Trieste

19/01/2023
SIGU Italian Medical Genetics Academy 2023 - I meeting

Online Webinar

27/01/2023 – 30/01/2023
SIGU "Academy Winter School"

Vipiteno

16/02/2023
SIGU - Italian Medical Genetics Academy 2023 - II meeting

Online Webinar

07/03/2023
Ethics Research

University of Trieste

09/03/2023
I seminari della Genetica: Displasie scheletriche e Il neonato piccolo per età gestazionale

University of Trieste - I.R.C.C.S. "Burlo Garofolo" - Trieste

16/03/2023
SIGU - Italian Medical Genetics Academy 2023 - III meeting

Online Webinar

20/03/2023
I seminari della genetica: Large scale biobank studies as tools to uncover the genetic background of diseases

University of Trieste - I.R.C.C.S. "Burlo Garofolo" - Trieste

13/04/2023
SIGU - Italian Medical Genetics Academy 2023 - IV meeting

Online Webinar

18/05/2023
SIGU - Italian Medical Genetics Academy 2023 - V meeting

Online Webinar

15/06/2023
SIGU - Italian Medical Genetics Academy 2023 - VI meeting

Online Webinar

13/07/2023
SIGU - Italian Medical Genetics Academy 2023 - VII meeting

Online Webinar

15/09/2023
SIGU - Italian Medical Genetics Academy 2023 - VIII meeting

Online Webinar

19/10/2023
SIGU - Italian Medical Genetics Academy 2023 - IX meeting

Online Webinar

16/11/2023
SIGU - Italian Medical Genetics Academy 2023 - X meeting

Online Webinar

04/12/2023
I seminari della Genetica

University of Trieste - I.R.C.C.S. "Burlo Garofolo" - Trieste
1. Prof. Han Brunner: “Why do we have the diseases we have?”
2. Prof. Alfredo Brusco: "Genetic Architecture of Autism Spectrum Disorders"
3. Prof. Massimo Gennarelli: "Genetics and Genomics in Clinical Psychiatry"

14/12/2023
SIGU - Italian Medical Genetics Academy 2023 - XI meeting

Online Webinar

08/01/2024 – 31/12/2024
IRCBG_24035 Presentazione di casi clinici complessi: risultati, analisi e discussione

I.R.C.C.S. "Burlo Garofolo" - Trieste

26/01/2024 – 29/01/2024
SIGU "Academy Winter School"

Vipiteno

05/02/2024
I seminari della Genetica

University of Trieste - I.R.C.C.S. "Burlo Garofolo" - Trieste
1. “Mutazioni rare e alleli comuni: effetti sulla valutazione del rischio genomico in malattie complesse e multifattoriali” – G. Novelli
2. “Research and diagnostics of rare diseases in Czechia” – M. Macek
3. “Clinical and molecular charaacterization of rare disease” – G. B. Ferrero

13/03/2024
Public speaking

University of Trieste

14/03/2024
I seminari della Genetica

“Development and plasticity of food preferences at the start of life” – L. Marlier

17/07/2024
Formazione generale dei Lavoratori

Online course

16/08/2024
IRCBG_24012 Prevenzione Delle Infezioni Ospedaliere (ICA). Modulo A (PNRR M6C2 2.2B)

Online course

11/10/2024
ASUGI_01850 1st International Meeting. Sudden Cardiac Death in the Balkan Area: beyond what we know and directions for the future research

Azienda sanitaria universitaria Giuliano Isontina

12/11/2024
IRCBG_24002 BLSD (Basic Life Support - Defibrillation) Sanitari Secondo Linee Guida ILCOR 2020

I.R.C.C.S. "Burlo Garofolo" - Trieste

18/11/2024
IRCBG_24001 Il Fascicolo Sanitario Elettronico (FSE) 2.0

I.R.C.C.S. "Burlo Garofolo" - Trieste

26/11/2024
IRCBG_24086 Trattamento e protezione dei dati personali

I.R.C.C.S. "Burlo Garofolo" - Trieste

24/01/2025 – 27/01/2025
SIGU "Academy Winter School"

Vipiteno

20/02/2025 – 21/02/2025
IRCBG_25007 Corso di formazione specifica per lavoratori per i settori della classe di rischio alto

I.R.C.C.S. "Burlo Garofolo" - Trieste

28/02/2025
Malattie Rare: Screening, Diagnosi e Terapie Innovative

I.R.C.C.S. "Burlo Garofolo" - Trieste

26/03/2025
IRCBG_25035 La rilevazione delle presenze dei dipendenti pubblici

I.R.C.C.S. "Burlo Garofolo" - Trieste

24/10/2025
Italian Cardiogenetics Group

Fondazione I.R.C.C.S. Policlinico San Matteo - Pavia

HONOURS AND AWARDS

12/09/2022
Spoendlin Junior Award – Symposium & 57th Inner Ear Biology Workshop – IEB 2022

Award for the presentation entitled "Dual molecular diagnosis in complex hearing loss patients: when a single gene is not enough".

04/10/2024
Best oral communication Award – XXVII SIGU National Congress

Award for the presentation entitled "A race against time: when high-throughput sequencing technologies are essential to provide critically-ill children admitted to Neonatal and Pediatric Intensive Care Units (NICU/PICU) with a molecular diagnosis".

17/10/2025
Reviewers' Choice Abstract – American Society of Human Genetics annual meeting - ASHG 2025

Award for the presentation entitled "Running against the clock: application of Next Generation Sequencing (NGS) technologies to diagnose critically-ill children admitted to Neonatal and Pediatric Intensive Care Units (NICUs/PICUs)".

TEACHING ACTIVITY

10/2023 – CURRENT
Medical Genetics

She teaches Medical Genetics in the following degree programmes of the University of Trieste:
- "Dental Hygiene" (both in Trieste and Pordenone)
- “Health Care Assistance”
- "Dietistic"

10/2023 – 09/2024
Medical Genetics

She has taught Medical Genetics in the degree programme "Psychiatric Rehabilitation Techniques" of the University of Trieste.

21/07/2022
Corsi di preparazione agli esami di ammissione d’area medica

Medical Genetics lesson during the preparation course for Medicine admission test.

01/12/2022 – CURRENT
Collaboration for the organisation of the seminars of the Specialisation School of Medical Genetics of the University of Trieste

1. Displasie scheletriche - dott. Luigi Memo - 09/03/2023
2. Il neonato piccolo per età gestazionale - dott. Gioacchino Scarano - 09/03/2023
3. Large scale biobank studies as tools to uncover the genetic background of diseases - Prof. Aarno Palotie - 20/03/2023

THESES CO-SUPERVISOR

2019
"The sense of Smell: Genomic studies of Human Knockouts”

Master degree in Medical Biotechnology - University of Trieste

2022
“Whole Genome Sequencing analysis and olfactory dysfunction: deep characterization of a highly selected cohort of COVID - 19 patients"

Master degree in Functional Genomics - University of Trieste

2023
"Empowering critical neonatal care: the impact of Whole Exome Sequencing in urgent diagnoses"

6-year Single-Cycle Degree in Medicine and Surgery - University of Trieste

2024
Beyond the curtains: the application of WES in fetuses after termination of pregnancy

6-year Single-Cycle Degree in Medicine and Surgery - University of Trieste

THESES EXTERNAL REFEREE

2022
"High throughput sequencing technologies: a large study on hereditary hearing loss patients"

Master degree in Functional Genomics - University of Trieste

2023
"Whole Exome Sequencing for the analysis of highly selected patients affected by Epileptic Encephalopathies (EEs) and Developmental and Epileptic Encephalopathies (DEEs)"

International Master degree in Neuroscience - University of Trieste

2023
"Caratterizzazione di mutazioni somatiche associate alla progressione della steatosi epatica a cirrosi ed epatocarcinoma"

Master degree in Medical and Diagnostic Biotechnologies - University of Trieste

2024
"The challenge in the diagnosis of malformed fetuses: genomic analysis and functional characterization of splicing variants"

Master degree in Medical and Diagnostic Biotechnologies - University of Trieste

2025
Shedding Light on Genetics of Dietary Intake: A Genome-Wide Association Study on a Friuli-Venezia Giulia Cohort

Master degree in Medical and Diagnostic Biotechnologies - University of Trieste

2025
Decoding Endometriosis: Exploring Rare Genetic Variants Using Whole Exome Sequencing and Assessing the Impact of Dietary Habits on Symptoms Management

Master degree in Medical and Diagnostic Biotechnologies - University of Trieste

NETWORKS AND MEMBERSHIPS

06/2018 – CURRENT Ordine dei Medici Chirurghi e Odontoiatri della provincia di Trieste
Young doctors committee

The Young doctors commitee is dedicated to working on issues and challenges facing junior doctors. This includes helping young colleagues with possible problems in entering the working world or accessing the different Specialisation schools after achieving the Master's degree in Medicine and Surgery. Moreover, the board mediates the relationship between young doctors and the Institutions, primarily the Medical Board.

28/03/2022 – CURRENT
Member of Società Italiana di Genetica Umana (SIGU)

2024 – CURRENT
Member of the "Comitato d'Indirizzo del Corso di Laurea Magistrale di Medicina e Chirurgia"

LANGUAGE SKILLS

Mother tongue(s): **ITALIAN**

Other language(s):

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken production	Spoken interaction	
ENGLISH	C1	C1	C1	C1	C1

Levels: A1 and A2: Basic user; B1 and B2: Independent user; C1 and C2: Proficient user

DRIVING LICENCE

Driving Licence: B | 13/10/2020 – 04/07/2031

Trieste , 25/10/2025