Beatrice Spedicati

Gender: Female **Email address:** beatrice.spedicati@burlo.trieste.it

WORK EXPERIENCE

Master scholarship

I.R.C.C.S. "Burlo Garofolo" [01/02/2023 – Current]

City: Trieste

Country: Italy

5x1000 2015 Project "Senses - Genetics of senses and related diseases"; CUP:C92F17003560001.
Her research work, carried out alongside her PhD program in Reproduction and Developmental Sciences, 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.

EDUCATION AND TRAINING

PhD Program in Reproduction and Developmental Sciences

Department of Medicine, Surgery and Health Sciences, University of Trieste [01/11/2021 – Current]

Address: Strada di Fiume, 447, 34149 Trieste (Italy)

Website: https://dsm.units.it/

She is currently participating in several research projects focusing primarily on the genetic of senses, i.e. hearing, smell, taste and sight. In particular, she is 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 is also taking part in other research projects, as the identification of the genetic bases of endometriosis and the genetics of odontostomatological traits. Additionally, she is expanding her research projects on the clinical and genetic characterisation of “Human Knockouts” and on dual molecular diagnoses in complex patients.

Specialisation School in Medical Genetics

Department of Medicine, Surgery and Health Sciences, University of Trieste [01/11/2018 – 30/10/2022]

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

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.

State exam - Professional register admmission

Department of Medicine, Surgery and Health Sciences, University of Trieste [11/2017 – 02/2018]

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

Master's Degree in Medicine and Surgery

Department of Medicine, Surgery and Health Sciences, University of Trieste [10/2011 – 10/2017]

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

Master's Degree Internship

International Centre for Genetic Engineering and Biotechnology - Molecular Medicine Group [07/2016 – 10/2017]

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

- 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

Degree in Organ and Organ Composition

"G. Tartini" State Conservatory of Trieste [11/2004 – 06/2016]

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

Secondary School Diploma

"G. Oberdan" Scientific High School [09/2006 – 07/2011]

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

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

[10/09/2022 – 13/09/2022]

"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/

Genomics of Rare Disease (Online conference)

[22/03/2021 – 24/03/2021]

"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/

XXII SIGU National Congress (Rome, Italy)

[13/11/2019 – 16/11/2019]

"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/

NATIONAL AND INTERNATIONAL CONFERENCES AND SEMINARS - POSTER PRESENTATIONS

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

[10/09/2022 – 13/09/2022]

"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/

XXV SIGU National Congress (Trieste, Italy)

[07/09/2022 – 09/09/2022]

"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/

XXV SIGU National Congress (Trieste, Italy)

[07/09/2022 – 09/09/2022]

"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/

European Human Genetics Conference (Hybrid converence Vienna - Virtual)

[11/06/2022 – 14/06/2022]

"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/

XXIV SIGU National Congress (Virtual conference)

[17/11/2021 – 19/11/2021]

"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/

European Human Genetics Conference (Virtual conference)

[28/08/2021 – 31/08/2021]

"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/

XXIII SIGU National Congress (Virtual conference)

[11/11/2020 – 13/11/2020]

"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/

European Human Genetics Conference (Virtual conference)

[06/06/2020 – 09/06/2020]

"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/

PARTICIPATION TO CONFERENCES, SEMINARS AND MEETINGS

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

[20/03/2023]

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

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

Online Webinar

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

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

Ethics Research
[07/03/2023]

University of Trieste

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

Online Webinar

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

Vipiteno

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

Online Webinar

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

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

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

University of Trieste

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

University of Trieste

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

University of Trieste

The sixth taste
[11/04/2022]

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

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

University of Trieste

Webinar on Cardiomyopathies
[30/03/2021]

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

Discussione di casi clinici e diagnostici complessi in genetica medica

[24/02/2020 – 28/02/2021]

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

BLSD for healthcare providers course

[02/2020]

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

New frontiers in research, diagnostics and therapies

[12/12/2019 – 13/12/2019]

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

Sindromi da iperaccrescimento: pathway e clinica

[15/05/2019]

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

Diagnosi prenatale: processi di integrazione multiprofessionale

[01/02/2019 – 31/12/2019]

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

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

[24/03/2018]

Ordine dei Medici Chirurghi e Odontoiatri della provincia di Trieste

PUBLICATIONS

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

[2023]

https://doi.org/10.3390/foods12091739

Authors:

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

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

[2023]

https://doi.org/10.3390/jcm12072746

Authors:

Valentina Luppieri, Alessandro Pecori, **Beatrice Spedicati**, Riccardo Schito, Lucia Pozzan, Aurora Santin, Giorgia Girotto, Milena Cadenaro, Maria Pina Concas

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

[2023]

https://doi.org/10.3390/biomedicines11030703

Authors:

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

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

[2023]

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

Authors:

Stefania Lenarduzzi, **Beatrice Spedicati**, Beatrice Alessandrini, Paola Tesolin, Alessia Paldino, Marta Gigli, Gianfranco Sinagra, Paolo Gasparini, Matteo Dal Ferro, Giorgia Girotto

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

[2022]

https://doi.org/10.3390/genes13112023

Authors:

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

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

[2022]

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

Authors:

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

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

[2022]

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

Authors:

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

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

[2022]

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

Authors:

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

Infant with a big head and 'crossed' polysyndactyly

[2022]

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

Authors:

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

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

[2021]

Authors:

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

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

[2021]

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

Authors:

Beatrice Spedicati, Massimiliano Cocca, Roberto Palmisano, Flavio Faletra, Caterina Barbieri, Margherita Francescatto, Massimo Mezzavilla, Anna Morgan, Giulia Pelliccione, Paolo Gasparini, Giorgia Girotto

The Role of Knockout Olfactory Receptor Genes in Odor Discrimination

[2021]

https://doi.org/10.3390/genes12050631

Authors:

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

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

[2020]

https://doi.org/10.3390/genes11111237

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

ABSTRACTS SUBMITTED TO NATIONAL AND INTERNATIONAL CONFERENCES AND MEETINGS

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

[2023]

Abstract submitted to Pangborn Sensory Science Symposium 2023

Authors:

C. A-M. Graham, **B. Spedicati**, G. Pelliccione, P. Gasparini, M. P. Concas

Unveiling the genetic bases of Hereditary Hearin Loss (HHL): the application of a multistep diagnostic approach in a large Italian cohort

[2023]

Abstract submitted to the European Human Genetics Conference 2023

Authors:

B. Spedicati, A. Santin, G. G. Nardone, S. Lenarduzzi, E. Rubinato, C. Graziano, L. Garavelli, S. Miccoli, S. Bigoni, A. Morgan, G. Girotto

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

[2023]

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

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

[2023]

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

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

[2023]

Abstract submitted to the European Human Genetics Conference 2023

Authors:

G. G. Nardone, A. Santin, A. Morgan, **B. Spedicati**, M. P. Concas, G. Girotto

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

[2023]

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

Genetic diseases in the arts

[2022]

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

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

[2022]

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

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

[2022]

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

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

[2022]

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

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

[2022]

Abstract submitted to the XXV SIGU National Congress

Authors:

A. Pecori, V. Luppieri, **B. Spedicati**, R. Schito, M. Cadenaro, G. Girotto, M. P. Concas

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

[2022]

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

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

[2022]

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

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

[2022]

Abstract submitted to the XXV SIGU National Congress

Authors:

E. Rubinato, A. Morgan, F. Faletra, A. Feresin, **B. Spedicati**, G. Girotto

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

[2022]

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

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

[2022]

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

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

[2022]

Abstract submitted to the European Human Genetics Conference 2022

Authors:

A. Morgan, S. Lenarduzzi, **B. Spedicati**, P. Tesolin, A. Santin, E. Rubinato, G. Girotto

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

[2022]

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

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

[2021]

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

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

[2021]

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

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

[2021]

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

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

[2021]

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

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

[2021]

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.

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.

[2021]

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

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

[2021]

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

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

[2021]

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

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.

[2021]

Abstract submitted to the Association for Research in Otolaryngology - Midwinter meeting 2021

Authors:

A. Morgan Anna, 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.

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

[2021]

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

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)

[2020]

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.

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

[2020]

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

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

[2020]

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

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

[2019]

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

HONOURS AND AWARDS

Spoendlin Junior Award

Symposium & 57th Inner Ear Biology Workshop – IEB 2022 [12/09/2022]

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

TEACHING ACTIVITY

Corsi di preparazione agli esami di ammissione d’area medica

[21/07/2022]

Medical Genetics lesson during the preparation course for Medicine admission test

Collaboration for the organisation of the seminars of the Specialisation School of Medical Genetics of the University of Trieste

[01/12/2022 – Current]

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

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

[2019]

Master degree in Medical Biotechnology - University of Trieste

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

[2022]

Master degree in Functional Genomics - University of Trieste

THESES EXTERNAL REFEREE

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

[2022]

Master degree in Functional Genomics - University of Trieste

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

[2023]

International Master degree in Neuroscience - University of Trieste

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

[2023]

Master degree in Medical and Diagnostic Biotechnologies - University of Trieste

NETWORKS AND MEMBERSHIPS

Member of Società Italiana di Genetica Umana (SIGU)

[28/03/2022 – Current]

Young doctors committee

[Ordine dei Medici Chirurghi e Odontoiatri della provincia di Trieste, 06/2018 – Current]

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.

LANGUAGE SKILLS

Mother tongue(s): **Italian**

Other language(s):

English

LISTENING C1 **READING** C1 **WRITING** C1

SPOKEN PRODUCTION C1 **SPOKEN INTERACTION** C1

DRIVING LICENCE

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

Curriculum ai fini della diffusione online

Trieste, 09/05/2023