CURRICULUM VITAE

PERSONAL INFORMATION

Surname and Name	**GIROTTO GIORGIA**
Professional qualification	**FULL PROFESSOR / PHD, RESIDENCY SCHOOL IN MEDICAL GENETICS**
Current work position	**FULL PROFESSOR / BIOLOGIST SPECIALISED IN MEDICAL GENETICS**
Phone contact	**+39 0403785538**
E-mail contact	**giorgia.girotto@burlo.trieste.it; ggirotto@units.it**

Educational and Professional Qualifications

2024: Nominated **Member of the Collegium Oto-Rhino-Laryngologicum Amicitiae Sacrum.**
2021: **National academic qualification in Medical Genetics "Professore di prima fascia"** (06/A1 Genetica Medica, BANDO D.D. 2175/2018**).**
2017: **National academic qualification in Medical Genetics "Professore di seconda fascia"** (06/A1 Genetica Medica, Bando D.D 1532/2016).
2017: **Medical Genetics Residency School in Medical Genetics** at the University of Genoa, Italy (4 years course).
2013: Registration in the National Biology Association (AA_067918).
2012: **PhD in Molecular Genetics with the award of Doctor Europaeus Mention,** PhD course in Reproduction and Developmental sciences**,** University of Trieste, Italy (3 years course).
2012: Board certification in Biology.
2009: **Master's Degree in Medical Biotechnology,** University of Trieste, Italy**.**
2006: **Biotechnology degree (Bachelor's Degree),** University of Trieste, Italy.
2003: **Secondary school diploma in Classical studies,** Mestre-Venice, Italy.

Current work experience

From October 2025: Full Professor of Medical Genetics at the Department of Medicine, Surgery and Health Sciences, University of Trieste.
From November 2024: Director of the Specialization School of Medical Genetics at the Department of Medicine, Surgery and Health Sciences, University of Trieste.
From September 2023: Director of the SS Laboratory of Genomics and Bioinformatics within the SC Medical Genetics unit at the Institute for Maternal and Child Health IRCCS Burlo Garofolo.
From October 2021 to October 2025: Associate Professor of Medical Genetics at the Department of Medicine, Surgery and Health Sciences, University of Trieste.
From January 2018: Biologist at Medical Genetics Units, Department of Services and Advanced Diagnostics of the Institute for Maternal and Child Health IRCCS Burlo Garofolo.

She actively participates in internal and external multidisciplinary training courses with particular reference to genomics and bioinformatics. She is personally involved in the organization of the diagnostic-clinical process for managing the genetic and genomic data analysis of the SC Genetica Medica patients.
The positive results of the evaluation of the diagnostic activity (valutazioni Dirigenti ai sensi dell'art. 59 del CCNL Area Sanità del 19/12/2019) at the SC Genetica Medica reported that:

1) She has completed the assignments entrusted to her, demonstrating considerable skills supported by extensive scientific activity (see abstracts and publications sections) as well as national and international grants won (as PI) focused on hereditary and multifactorial diseases and their study through genetic/genomics analyses.
2) She has actively participated in laboratory control procedures and quality

control of clinical services.

3) She has implemented and promoted technological innovations (with particular reference to the field of genomics and bioinformatics), allowing to a) centralize genetic analysis of some pathologies (e.g., hearing loss, cardiomyopathies, malformation syndromes, critically ill infants), thus making the Institute regional and national reference center, b) reducing the time of analysis and improving the molecular diagnostic path.
4) She is member of several working groups belonging to the Italian Society of Human Genetics (e.g. GdL -omics and cytogenomics sciences, multifactorial diseases), and national groups focused on complex malformative syndromes (Rete IDEA), and cardiology and otorhinolaryngology groups.
5) She has organized numerous national and international conferences in the field of genetics and genomics as being Professor of various university courses at the Department of Medicine, Surgery and Health Sciences, as well as of the Doctoral School in Reproduction and Developmental Sciences, and Specialization School in Medical Genetics.

Research activity

- Coordinator of a research group focused on:
    - Hearing/auditory function and loss, sensorial traits (e.g., smell, taste, sight), and correlated pathologies, with the aim of identifying genes involved in and related to sensory function and decay.
    - Complex diseases, such as endometriosis, psychiatric traits, odontostomatological traits and cardiometabolic diseases.
    - Clinical and genetic characterization of "Human Knockout" in genetically isolated populations.
- Coordinator of the international G-EAR Consortium, whose activity aims at further understand the genetics of hearing function and hearing loss.
- Member of more than 20 internationals consortia for studying complex traits and diseases through GWAS studies, supporting their samples collection and data analysis.

Diagnostic activity:

Referent for the coordination and analysis of NGS data on hereditary diseases, such as hearing loss (syndromic and non-syndromic forms), cardiomyopathies, and other malformative syndromic cases. She also carries out pre-test genetic counselling for several hereditary diseases, as well as genetic counselling for hearing loss.

Past work experience

- 2019-2021: **Researcher (RTDB),** Department of Medicine, Surgery and Health Sciences, University of Trieste. During this period, she strengthened the long-standing national and international collaborations, further pursuing her diagnostic and research activities.
- 2015-2019: **Researcher (RTDA**), Department of Medicine, Surgery and Health Sciences, University of Trieste. During this period, she strengthened the long-standing collaboration with the Qatari geneticists and audiologists. In particular, she has been involved in the collaboration with Hamad Medical Corporation (HMC) (Doha, Qatar) but also with Sidra Medical Reseach Hospital regarding the project entitled "Genetics of Deafness in the Qatari Population". For this project she applied Linkage analysis and Next Generation Sequencing technology to analyse Qatari families affected by hereditary hearing loss. Thanks to this collaboration she visited several times the HMC and, from August 2015 to November 2016, she was Visiting Scientist at Sidra Medical Hospital, performing analyses, delivering speeches and organizing some training activities with the local researchers.
- 2013-2015: **Research Fellow**, Department of Medicine, Surgery and Health Sciences, University of Trieste. She was involved in a large project aimed at the identification of the molecular bases of hearing loss (both congenital and late-onset forms), as well as the coordination of the International Consortium G-EAR. During this period, she spent two months as Visiting Scientist at the Centre for Genomic Regulation (CRG) (Barcelona, Spain) to run bioinformatics and biostatistics analyses of whole-exome sequencing data. She also spent six months as Visiting Scientist at the Mary Lyon Centre (Harwell, UK) to perform experiments on mice

models for a collaborative project on age-related hearing loss. Moreover, she was an organizer and a participant in a pilot scientific expedition to collect DNA samples, together with audiological and taste phenotypes, in a remote area of Mongolia.

- 2010-2012: **PhD student** in the PhD Program in Molecular Genetics with the award of Doctor Europaeus Mention (PhD course in Reproduction and Developmental Sciences), Department of Medicine, Surgery and Health Sciences, University of Trieste. During her PhD program, she has started the coordination of the International Consortium G-EAR focused on identifying molecular bases of presbycusis through a metanalysis of GWAS data (mainly coming from isolated populations). During the second year of her PhD, she spent six months as Visiting Scientist at the Wellcome Trust Sanger Insitute (Cambridge, UK) in the "Genetics of Deafness laboratory". Moreover, she partecipated in the "Marco Polo Scientific Expedition", aimed at the collection of samples and execution of audiometric test along the Silk Road (Georgia, Azerbajan, Turkmenistan, Tajikistan, Crimea, Uzbekistan, Kazakhstan, Kirghizistan, China).
- 2009: **Short-term Fellowship** at the Medical Genetics Unit of IRCCS Burlo Garofolo.
- 2009: **Visiting Scientist** at the National Institute for Medical Research (London, UK) in the Molecular Structure laboratory.

Teaching activity and student's evaluation

She teaches Medical Genetics in the following courses:

- "Medicine and Surgery" (786ME-2) from 2022, University of Trieste. Average of student's evaluation: 8.22 (academic year 2022-2023) , 8.03 (academic year 2023-2024)
- "Dentistry" (969ME-2) from 2022, University of Trieste. Average of student's evaluation: 7.98 (academic year 2022-2023), 7.35 (academic year 2023-2024).
- "Speech and Language Therapy" (419ME-3) from 2022 to 2024, University of Trieste. Average of student's evaluation: 7.56 (academic year 2022-2023), 8.44 (academic year 2023-2024).
- "Diagnostic and Medical Biotechnologies" (983SV) from 2021 to 2022 and from 2023, University of Trieste. Average of student's evaluation: 7.13 (academic year 2021-2022), 8.42 (academic year 2023-2024).
- "Health Care Assistance" (692ME-3) from 2021 to 2022, University of Trieste. Average of student's evaluation: 8.45 (academic year 2021-2022).
- "Psychiatric Rehabilitation Techniques" (367ME-3) from 2019 to 2023, University of Trieste. Average of student's evaluation: 8.24 (academic year 2019-2020), 8.38 (academic year 2020-2021), 8.28 (academic year 2021-2022), 8.19 (academic year 2022-2023).
- "Psychology" from 2018 to 2019, University of Trieste.
- "Dental Hygiene" (025ME-1) from 2019 to 2023, University of Trieste. Average of student's evaluation: 8.76 (academic year 2019-2020), 8.06 (academic year 2020-2021), 8.19 (academic year 2021-2022), 9.21 (academic year 2022-2023).

She teaches Innovative Therapies in Medical Genetics (744ME) in the following courses:

- "Medicine and Surgery" from 2019 to 2024, University of Trieste. Average of student's evaluation: 8.55 (academic year 2019-2020), 7.83 (academic year 2020-2021), 9.22 (academic year 2021-2022), 7.90 (academic year 2022-2023), 9.64 (academic year 2023-2024).

She teaches "Omics Technologies in Medical Genetics" (976SV) in the following courses:

- "Diagnostic and Medical Biotechnologies" from 2022 to 2023, University of Trieste. Average of student's evaluation: 8.21 (academic year 2022-2023).
- "Functional Genomics" from 2022 to 2023, University of Trieste. Average of student's evaluation: 6.28 (academic year 2022-2023).

She also teaches in:

- Doctoral Programme in "Personalised Medicine and Innovative Therapies" from 2018, University of Trieste.
- Specialization School in "Medical Genetics" (SSM18 and SSM22) from 2016,

University of Trieste.
- Specialization School in “Gynecology and Obstetrics” (SSM06) from 2019 to 2021, University of Trieste.
- Specialization School in “Endocrinology and Metabolic Disorders” (SSM25) from 2022, University of Trieste.
- Specialization School in “Paediatric orthodontics” (SSO02) dal 2022, from 2022, University of Trieste
- II level Master in “Implantologia protesica uditiva” from 2016 to 2021, “Sapienza” University of Rome.
- Master in “Chirurgia oncoplastica della mammella” from 2021, University of Trieste.
- Master in “Basic science in Audiology” in 2021 University of Turin and Padua.
- Master in “Scienze uditive dell’età evolutive” in 2021, University of Perugia.
- Master in “Basic science in audiologia e foniatria” in 2022, Centro ricerche e studi Amplifon.

Supervisor and co-supervisor of theses:
To date, she supervised Bachelor, Master, Specialization and Doctoral degrees theses, as follows:
- 2022/2023: “Empowering critical neonatal care: the impact of Whole Exome Sequencing in urgent diagnoses”.
- 2021/2022: “Caratterizzazione di mutazioni somatiche associate alla progressione della statosi epatica a cirrosi ed epatocarcinoma”.
- 2021/2022: “Whole Exome Sequencing for the analysis of highly selected patients affected by Epileptic Encephalopathies and Developmental and Epileptic Encephalopathies”.
- 2021/2022: “High Throughput sequence technologies: a large study on hereditary hearing loss patients”.
- 2021/2022: “Endometriosis disease: novel genetic insights on a deeply clinical characterized Italian cohort”.
- 2021/2022: “Whole Genome Sequencing analysis and olfactory dysfunction: deep characterization of a highly selected cohort of COVID-19 patients”.
- 2020/2021: “Behind the scenes of complex models of inheritance: dual molecular diagnoses explain entangled clinical pictures”.
- 2020/2021: “Caratterizzazione genetica delle cardiomiopatie attraverso approcci di sequenziamento di nuova generazione”.
- 2020/2021: “Nuove scoperte sulla genetica della sindrome di Pendred: l'efficacia delle tecnologie di sequenziamento ad alto processamento e di una profonda valutazione clinica”.
- 2020/2021: “Genetic dissection of Cloninger’s Temperament and Character Inventory in Italian isolates”.
- 2020/2021: “Genetics bases of color vision defects in Silk Road isolated communities”.
- 2019/2020: “Applicazioni cliniche del sequenziamento dell'intero esoma: identificazione di diagnosi molecolari doppie in pazienti affetti da ipoacusia”.
- 2019/2020: “Identificazione e caratterizzazione funzionale di nuovi geni candidati e varianti associate a Sordità Ereditaria e Presbiacusia”.
- 2019/2020: “Human Knockouts e analisi fenotipica negli isolati genetici”.
- 2018/2019: “The sense of Smell: Genomic studies of Human Knockouts”.
- 2018/2019: “High-throughput data analysis of hearing phenotypes on 9000 subjects from ten cohorts and in 200.000 individuals from UK Biobank”.
- 2018/2019: “Studi di genomica: il ruolo degli Human Knockouts nell'era della medicina di precisione”.
- 2017/2018: “Tecnologie di sequenziamento di nuova generazione per la caratterizzazione molecolare di casi sindromici con fenotipo uditivo”.
- 2016/2017: “Age related hearing loss: in vitro and in vivo studies to characterize new candidate genes”.
- 2016/2017: “Studio prospettico della funzione uditiva e della presbiacusia in popolazioni geneticamente isolate”.
- 2015/2016: “Identification of New Hereditary Hearing Loss Genes Using High-Throughput Sequencing Technologies”.

- 2013/2014: “Usher syndrome: clinical features and molecular analysis using targeted resequencing - Sindrome di Usher: aspetti clinici e molecolari con utilizzo di tecnologia targeted resequencing”.

Other accademic assignments

- Member of the Board from 2018 of the Doctoral Programme in “Personalised Medicine and Innovative Therapies” , Department of Medicine, Surgery and Health Sciences, University of Trieste. Organizer of the “PhD Week”.
- Member of the Board of the Medical Genetics residency school (SSM18 and SSM22) from 2018 at the Department of Medicine, Surgery and Health Sciences, University of Trieste
- Member of the Thesis Defence committee in several graduation sessions for the following degree courses: Medicine, Biology, Biotechnology, Medical Biotechnology, and Functional Genomics.
- Delegate with responsibility for Linguistic from 2017 to 2021 at the Department of Medicine, Surgery and Health Sciences, University of Trieste
- Delegate for Research Commission from 2021 at the Department of Medicine, Surgery and Health Sciences, University of Trieste.
- Member of the CVR from May 2024 for the Department of Medical, Surgical and Health Sciences, University of Trieste.

Other assignments

- Organizer of a scientific expedition named “Geni Friulani nel Mondo, Brasile 2020
- Organizer of the international meeting on hearing loss “Symposium & 57th Inner Ear Biology Workshop – IEB 2022” (10-13/09/2022, Trieste, Italy).
- Member of the Scientific Board Cinzia Vitale Onlus, from 2022.
- Member of the following working groups of the Italian Society of Human Genetics (SIGU):
    - “Scienze omiche e citogenomica, genetica prenatale e riproduttiva”.
    - “Malattie Multifattoriali”.
- Guest Editor for a special issue of the scientific journal “Genes”, title: “Genetics and Epigenetics of Hearing Loss”, from 2020 to 2021.
- Guest Editor for a special issue of the scientific journal “Audiology Research” “Genetics of Hearing Loss—Volume II”. Deadline for manuscript submissions: 30 September 2023.
- Member of the Scientific Board of HUGO meeting 2024

Grants awarded

- 2015: Scientific Independence of Young Researchers (SIR) grant (RBSI14AG8P). Title of the project: “Age-related hearing loss: from gene identification to a better molecular diagnosis and prevention” (Role: PI).
- 2016: Junior Scientists Research Experience Program grant (JSREP07-013-3-006). Title of the project: “Age-related hearing loss in Qatar: a genomic approach to identify causative genes” (Role: PI).
- 2019: Beneficentia Stiftung foundation. Title of the project: “Hereditary Hearing Loss: new genes discovery for a preventive strategy and the development of new therapeutic targets” (Role: PI).
- 2019: MIUR project: “SENSAGING project “Sensory decays and ageing” (2019-2022) (Role: participant).
- 2020: Grant entiled “Hearing loss: from genes identification to personalized diagnosis and treatments” awarded to the Department of Medicine, Surgery and Health Sciences, University of Trieste (Role: PI).
- 2020: ENDO-2020-23670288 entitled “Endometrosis: Pathogenesis of endometriosis: the role of genes, inflammation and environment” awarded to IRCCS Burlo Garofolo, Trieste, Italy (Role: PI).
- 2021*:* D70-FRA-2021 (University of Trieste)
- 2021: RC-02-2021 project entitled “A unique cohort to study mechanisms underlying sex differences in cardiometabolic diseases”, awarded to IRCCS Burlo Garofolo, Trieste, Italy (Role: PI)
- 2021: RF-2021-12374963 project entitled “Less genes more genomes: modeling the implementation of integrative -OMICs as a first line tool in the clinical practice” awarded to Ospedale Pediatrico “Bambino Gesù”, Rome, Italy (Role: participant).

- 2022: PNRR-MR1-2022-12376811 project, entitled “Implementation of a standardized workflow for a more effective management and care of patients with syndromic and isolated intellectual disability” awarded to Ospedale Pediatrico “Bambino Gesù”, Rome, Italy (Role: participant).

National and International collaborations

- Participation in the INGI consortium on genetically isolated populations.
- International collaborations for the generation of animal models aimed at the study of hearing loss (King’s college London-UK; MRC-Harwell, UK; Harvard Medical School-Howard Hughes Medical Institute, Department of Neurobiology, Boston MA, United States; Harvard Medical School and Brigham and Women’s Hospital, Department of Pathology, Boston MA, United States).
- International collaboration for the study of hearing loss in Qatari population (Audiology and Balance Unit, and Molecular Genetics Laboratory and Laboratory of Medicine and Pathology, Hamad Medical Corporation, Doha, Qatar).
- National collaboration with colleagues affiliated to: University of Milan, Fondazione IRCCS Cà Granda (Milan, Italy), Ospedale Maggiore Policlinico (Milan, Italy), Catholic-University, Rome, University of Naples, University of Ferrara, Medical Genetic Units of Cesena and Monza, University of Bari, Hospital S.Orsola-Malpighi of Bologna, University of Ferrara, University of Padua, IRCCS De Bellis of Castellana Grotte, University of Insubria, University of Verona, University of Bologna, CNR, Cagliari Hospital.
- Participation in international consortia, including: ICBP (International Consortium Blood Pressure), CHARGE (Cohorts for Heart and Ageing Research in Genor Epidemiology), GIANT (The Genetic Investigation of Anthropometric Traits), TI Visigen Consortium, AlcGen Consortium, CDKGen Consortium, The Reprog consortium, CRP study, ALSPAC Consortium, CalciGen Consortium, Hum Reproductive Behaviour Consortium, NeuroCHARGE Consortium, Dr.Sud Seshadri (UT Health San Antonio), Dr. Galit Weinstein (Tel Aviv University).
- International collaboration with Professor Manuel Palacin for the identification molecules able to promote the expression of a gene involved in Hearing Lo (Institute for Research in Biomedicine, Barcelona, Spain).
- Participation to board of experts, in order to collaborate on the writing of nation guidelines for coclear implants.

Courses and conferences attended

- Participation to more than eight national and international courses since 2010 on genomic data for the study of monogenic and multifactorial diseases and the implementation of statistics and bioinformatics pipelines.
- Participation to more than 85 national and international conferences and workshops since 2009 on medical genetics, molecular biology, management and analysis of sequencing data, development and implementation of next-generation technologies for investigating the molecular causes of genetic diseases. In particular, Qatar Foundation Annual Research Forum (HMC Hospital, Qatar; 2010), International Conference on Complex Traits and Genetic Isolates (Arbatax, Italia; 2010), XIV SIGU Conference (Milan; 2011), 35th MidWinter Meeting ARO 2012 (San Diego, California; 2012), International meeting on complex traits and genetic isolates (Trieste, Italy; 2012 Mar 15-17), The Brain Prize Meeting (Middelfart, Denmark; 2012 Oct 29-30), XV SIGU Conference 2012 (Sorrento, Italy; 2012 Nov 21-22), 36th Annual MidWinter Meeting ARO 2013 (Baltimore, USA; 2013 Feb 16-20), European Human Genetics Conference 2013 (Paris, France, 2013 Jun 8-11), “Rock around the cochlea” session SIGU 2013 (Rome, Italy; 2013 Sep 25-28), 37th Annual MidWinter Meeting ARO 214 (San Diego, USA; 2014 Apr 22-26), European Human Genetics Conference 2014 (Milan, Italy; 2014 May 31-Jun 3), 51st Annual Conference Inner Ear Biology Workshop (Sheffield, UK; 2014 Aug 30 – Sep 2), ARC’14 (Doha, Qatar, 2014 Nov 18-19), 38th Annual MidWinter Meeting ARO 2015 (Baltimore, US; 2015 Feb 21-25), IEB 2015 (Rome, Italy; 2015 Sep 12-15), Genetika 2015 (Rogaška Slatina, Slovenia; 2015 Sep 20-23), 9th Annual MidWinter Meeting ARO 2016 (San Diego, USA; 2016 Feb 20-24), SIGU 2016 (Torino, Italy; 2016 Dec 23-26), MBHD 2018 (Gottingen, Germany; 2018 May 16-19), XXI Congresso Nazionale SIGU 2018 (Catania, Italy; 2018 Oct 25-27), XXII Congresso Nazionale SIGU 2019 (Rome, Italy; 2019 Nov 13-15), European Human Genetics Conference 2020 (Online 2020 June 06 - 09), Eurosense 2020 (Rotterdam, Netherlands; 2020 December 13-16), XXIII Congresso Nazionale SIGU 2020 (Online; 2020 Nov 11-

13), European Human Genetics Conference 2021 (Online; 2021 Ago 28 - 31), ARO Meeting 2021 (Online; 2021 Feb 20-24), XXIV Congresso Nazionale SIGU 2021 (Online; 2021 Nov 17-19), ARO Meeting 2022 (Online; 2022 Feb 05-09), XXV Congresso Nazionale SIGU 2022 (Trieste, Italy; 2022 Sep 07-09), IEB 2022 (Trieste, Italy; 2022 Sep 10-13), European Human Genetics Conference 2022 (Glasgow, UK; 2022 June 11 - 14), IEB 2023 (London, UK; 2023 Sep 2-5), EMBL PhD Symposium 2023 (Heidelburg, Germany; 2023 Nov 20-22), XXVI Congresso Nazionale SIGU 2023 (Rimini, Italy; 2023 Oct 04-06), ASHG 2023 (Washington, USA; 2023 Nov 01-05) Human Genome Meeting 2024 (Rome, Italy; 2024 Apr 08-10), European Human Genetics Conference 2024 (Berlin, Germany; 2024 June 01 - 04), Science Get Together 2024 (Nuremberg, Germany; 2024 June 28th), XXVII Congresso Nazionale SIGU 2024 (Padova, Italy; 2024 Oct 02-04), European Human Genetics Conference 2025 (Milan, Italy; 2025 May 24 - 27).

- Participation to more than 20 national and international conferences and workshops as invited speaker or chairman.
- Participation to more than 11 seminars and six master courses as invited lecturer.

Honours and awards

As first presenting author

- ESHG Poster Award at the European Meeting Conference 2011 (Amsterdam, The Netherlands, 2011) for the poster entitled: “New genes for normal hearing function and age-related hearing loss by genome-wide association and expression studies”.
- ESHG/SIGU Oral Presentation Award for the best oral presentation in clinical genetics in the memory of Claudio Castellan (Milan, Italy 2014). Title of the presentation: “New Hereditary hearing loss (HHL) genes/mutations identified by high throughput sequencing and genotyping in the Italian and Qatari populations”.

As co-author or PI of the work

- Qatar Foundation Annual Research Forum 2010 (Doha, Qatar). First prize in the Biomedicine category 2010 for the best oral presentation at the Qatar Foundation's first Annual Research Forum. Title of the presentation: “Mutations in GJB2, GJB6 and mDNA 1555 G>A explain only a minority of cases of Nonsyndromic Hearing Loss in the Qatari Population” (G. Girotto as co-author of the presentation done by Alkowari et al.).
- Inner Ear Biology, Spoendlin Junior Award 2015 (G. Girotto as PI of the work done by Anna Morgan). Title of the presentation: “A new targeted re-sequencing panel for unveiling the the genetic causes of age-related hearing loss (ARHL).
- ESHG Poster Award at the European Meeting Conference 2016 (Barcelona, Spain) (G. Girotto as PI of the work done by Vuckovic D.). Title of the poster: “Unravelling human complex traits: the case of hearing function and age related hearing loss”.
- Molecular Biology of Hearing and Deafness (MBHD) Award 2016. (G. Girotto as PI of the work done by Anna Morgan). Title of the presentation: “Genome Wide Association Studies (GWAS), Targeted Re-Sequencing (TRS) and Functional Studies: a powerful approach for the discovery of the genetic causes of Age-Related Hearing Loss (ARHL)”.
- ARO 39th Annual MidWinter Meeting Travel Award 2017 (G. Girotto as PI of the work done by Anna Morgan). Title of the presentation: "Genome Wide Association Studies (GWAS), Target- ed Re-sequencing (TRS) and Functional Validations for Unravelling Human Complex Traits: The Case of Hearing Function and Age Related Hearing Loss”
- SIGU Oral Communication Award 2020 (G. Girotto as PI of the work done by Anna Morgan) Title of the presentation: "Clinical application of Whole Exome Sequencing (WES): identification of dual molecular diagnoses in patients affected by Hearing Loss (HL)".
- SIGU Oral Communication Award 2021 (G. Girotto as PI of the work done by Anna Morgan) Title of the presentation: “Clinical application of Whole Exome Sequencing (WES): identification of dual molecular diagnoses in patients affected by Hearing Loss (HL)”.
- SIGU Oral Communication Award 2022 for the best oral communication in multifactorial diseases in the memory of Giuseppe Pilia (G. Girotto as PI of the work done by Aurora Santin) Title of the presentation: “Normal Hearing Function (NHF) and Age-Related Hearing Loss (ARHL): new candidate genes

identification from Genome-Wide Association Studies (GWAS) in Moli-sani cohort".

- Inner Ear Biology, Spoendlin Junior Award 2022 (G. Girotto as PI of the work done by Beatrice Spedicati). Title of the presentation:" Dual molecular diagnosis in complex hearing loss patients: when a single gene is not enough".
- ESHG Poster Award at the European Meeting Conference 2023 (G. Girotto as PI of the work done by Paola Tesolin). Title of the poster:" Repurposing drugs to treat SLC7A8 age-related hearing loss".
- SIGU Oral Communication Award 2024 for the best oral communication in multifactorial diseases in the memory of Giuseppe Pilia (G. Girotto as PI of the work done by Aurora Santin) Title of the presentation: "Age-Related Hearing Loss (ARHL) genetics at a corner: a GWAS meta-analysis on Italian cohorts".
- SIGU Oral Communication Award 2024 for the best oral communication (G. Girotto as PI of the work done by Beatrice Spedicati).

Books and chapters in books

- Scientific consultant for the printed and multimedial section of the book entitled "Futuri scienziati" and "Accademia delle scienze"- educational project for secondary schools (Flaccavento-Romano, Fabbri Editori 2012).
- Scientific consultant for educational project for printed and multimedial section of the book entitled "Futuri scienziati" and "Accademia delle scienze"- educational project for secondary schools (Flaccavento-Romano, Fabbri Editori 2013).
- Book Chapter: Dipresa S, Fabretto A, Girotto G, Zadro C, Gasparini P. "Genetics of Hearing Loss (from Congenital Forms to Presbycusis)". In: Dupont, JP, editor. Hearing loss: Classification, Causes and Treatment. Hauppauge, New York: Nova Science Publishers; 2011.
- Book Chapter: Ambrosetti U, Castorina P, Gasparini P, Girotto G. "Sordità da cause genetiche". In: Ambrosetti, U, editor. Audiologia protesica. Torino, Italy: Edizioni Minerva Medica; 2018.
- Book Chapter: Morgan A, Gasparini P, Girotto G. "Hearing loss". In: Lázaro C, Lerner-Ellis J, Spurdle, A., editors. "Clinical DNA Variant Interpretation: Theory and Practice" a book in the series "Translational and Applied Genomics". Amsterdam: Elsevier/Academic Press; 2021.
- Book Chapter: "Guida per cervelli affamati. Perché da bambini odiamo le verdure e altri misteri neurogastronomici che ci rendono umani". In: Coricelli C, Rossi SE, editors. Milano: Il Saggiatore; 2021.

Dissemination activity

Various interviews for local and national newspapers (e.g., Il Piccolo, 24oreNews. ilFriuli.it, triestecafe.it, Nordest21.it), radio and TV channels (e.g. Oggi Scienza TV, Rai Scienza, La Repubblica, Il Giornale dei Biologi, Il Bo Live UniPD, Mercurio Associazio Economica Italo-Tedesca, Rai FVG), including:

- 2020: Invited Speaker at the 7th meeting "Convivere ad Auschwitz" (Trieste, Italy)
- 2021: Speaker at "Trieste Next: spazio 13. Endometriosi, genetica e ambiente. Nuovi scenari per la diagnosi e la cura" (Trieste, Italy).
- 2021: Invited speaker at Premio Papa Enrnest Hemingway during the session "L'Approfondimento" (Caorle, Italy).
- 2022: Interview for "La Stampa - Tutto Scienze". Title: "Perché la tentazione del genoisimo è un'illusione pericolosa?".
- 2022: Interview for "Il Piccolo - Scienza".
- 2022: Speaker at "Trieste Science+Fiction Festival". Title of the presentation: "Eugenetica e genoismo: fiction o realtà?" (Trieste, Italy).
- 2023: Speaker at "The Experience of the Friuli Venezia Giulia Regional Register". Title presentation: "Empowering the application of the molecular autopsy in Sudden Cardiac Deaths (SCD)".
- 2024: Trieste Next: "ICONODIAGNOSTICA: L'INCONTRO DI ARTE E GENETICA" alla 13° edizione di Trieste Next "GLI ORIZZONTI DELL'INTELLIGENZA. La conoscenza e le frontiere tra essere umano e tecnologie" (Trieste, Italia).
- 2024: Interview on "Buongiorno Regione" RAI FVG "Women4Health Project".
- 2025: Interview on "Radar", RAI FVG "Conosci l'esposoma? Conseguenze del cambiamento climatico negli organismi."

Further, she participated to public events, including:

- Local social events.
- Sponsored sporting events.
- Fundraising and charity events.
- Science festivals.

Publications

ORCHID ID: 0000-0003-4507-6589
SCOPUS ID: 57221103405
H INDEX: 36

1. de Rocco D, Heller PG, Girotto G, Pastore A, Glembotsky AC, Marta RF, Bozzi V, Pecci A, Molinas FC, Savoia A. "MYH9 related disease: a novel missense Ala95Asp mutation of the MYH9 gene". Platelets. 2009 Dec;20(8):598-602. doi: 10.3109/09537100903349620, PMID: 19860543.
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- Pianigiani G, Morgan A, Rubinato E, Faletra F, Garavelli L, Gasparini P, Girotto G. "Analysis of patient-derived mRNA samples or minigene splicing assays as reliable and fast methods to validate novel variants potentially affecting splicing in the context of inherited diseases". SIGU 2022; Trieste, Italia (Poster presentation).
- Spedicati B, Morgan A, Bonati MT, Luglio A, Rubinato E, Suergiu S, Gasparini P, Faletra F, Girotto G. "Behind the scenes of entangled clinical pictures: the intriguing role of dual molecular diagnoses". SIGU 2022; Trieste, Italia (Poster presentation P130).
- Tesolin P, Roesch S, Dossena S, Zampollo S, Santin A, Rubinato E, Brotto D, Morgan A, Girotto G. "Next-generation sequencing technologies to molecularly diagnose patients affected by Pendred syndrome-like symptoms". SIGU 2022; Trieste, Italia (Poster presentation P122).
- Santin A, Tesolin P, Spedicati B, Nardone GG, Zito G, Romano F, Di Lorenzo G,

Morgan A, Concas MP, Ricci G, Girotto G. "Novel genetic insights into Endometriosis (EM) disease: the first Italian Whole-Exome sequencing (WES) study". SIGU 2022; Trieste, Italia (Poster presentation P099).

- Spedicati B, Nardone GG, Concas MP, Crudele F, Pecori A, Santin A, Tirelli G, Gasparini P, Morgan A, Boscolo-Rizzo P, Girotto G. "Unravelling the genetic bases of persistent olfactory dysfunction in COVID-19 patients: the psychophysical and molecular characterization of a large Italian cohort". SIGU 2022; Trieste, Italia (Poster presentation P096).
- Faletra F, Bonati MT, Morgan A, Rubinato E, Feresin A, Luglio A, Spedicati B, Girotto G, Gasparini P. "Whole Exome Sequencing in unravelling complex cases of bicuspid aortic valve". SIGU 2022; Trieste, Italia (Poster presentation P080).
- Rubinato E, Morgan A, Faletra F, Feresin A, Spedicati B, Girotto G. "The challenge of non-syndromic mimics: our experience with hereditary hearing loss". SIGU 2022; Trieste, Italia (Poster presentation P077).
- Rubinato E, Faletra F, Feresin A, Morgan A, Girotto G. "Confirmating the causative role of *SF3B2* in craniofacial microsomia: the first Italian family". SIGU 2022; Trieste, Italia (Poster presentation P076).
- Feresin A, Luglio A, Spedicati B, Morgan A, Bonati MT, Faletra F, Rubinato E, Stampalija T, Bussani R, Murru F, Girotto G, Gasparini P. "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". SIGU 2022; Trieste, Italia (Poster presentation P158).
- Pecori A, Luppieri V, Spedicati B, Schito R, Cadenaro M, Girotto G, Concas MP. "Orthodontic measurements in isolated populations from North-Eastern Italy: an epidemiological and genetics study". SIGU 2022; Trieste, Italia (Poster presentation).
- Alessandrini B, Spedicati B, Luglio A, Della Monica M, Zampino G, Scarano G, Gasparini P, Girotto G, Memo L. "The sense of the genetic diversity: a comparison between medicine and arts". SIGU 2022; Trieste, Italia (Podium presentation).
- Lenarduzzi S, Alessandrini A, Dal Ferro M, Paldino A, Sinagra G, Gasparini P, Girotto G. "Cardiomyopathies (CMs) and Whole-Exome Sequencing (WES): clinical characterization of an Italian cohort of patients". SIGU 2022; Trieste, Italia (Poster presentation).
- Santin A, Lenarduzzi S, Nardone GG, Spedicati B, Morgan A, Persichilli M, De Curtis A, Costanzo S, Gialluisi A, Iacoviello L, Gasparini P, Concas MP, Girotto G. "Deepening the genetics of hearing: Genome-wide Association Studies (GWAS) on Moli-sani cohort". IEB 2022; Trieste, Italia (Podium presentation id161).
- Spedicati B, Morgan A, Ambrosetti U, Garavelli L, Lenarudzzi S, Pelliccione G, Peluso F, Santin A, Gasparini P, Girotto G. "Dual molecular diagnosis in complex hearing loss patients: when a single gene is not enough". IEB 2022; Trieste, Italia (Podium presentation id131).
- Tesolin P, Morgan A, Pianigian G, Bartoccioni P, Nunes Martinez V, Palacin M, Girotto G. "Age-related hearing loss and *SLC7A8*: identification of molecules able to restore the gene lost activity". IEB 2022; Trieste, Italia (Podium presentation id126).
- Spedicati B, Morgan A, Garavelli L, Nardone GG, Pelliccione G, Pianigiani G, Gasparini G, Girotto G. "There's more behind Hereditary Hearing Loss: molecular and phenotypic expansion of *PPP1R12A*-related disorder". IEB 2022; Trieste, Italia (Poster presentation).
- Rubinato E, Morgan A, Faletra F, Feresin A, Spedicati B, Girotto G. "Non-Syndromic Or Syndromic Hearing Loss? Our Experience with The Challenge Of Non-Syndromic Mimics". IEB 2022, Trieste, Italia (Poster presentation).
- Pianigiani G, Morgan A, Garavelli L, Gasparini P, Girotto G. "Syndromic and non-syndromic hearing loss: identification and functional characterization of putative novel splicing variants". IEB 2022, Trieste, Italia (Poster presentation).
- Nardone GG, Crudele F, Santin A, Morgan A, Concas MP, Girotto G. "Overcoming the limits of Whole Exome Sequencing: the application of Whole Genome Sequencing for the molecular diagnosis of Hearing Loss". IEB 2022; Trieste, Italia (Poster presentation).
- Tesolin P, Roesch S, Dossena S, Zampollo S, Rubinato E, Brotto D, Morgan A, Girotto G. "Pendred syndrome and related phenotypes: the definition of a

molecular diagnosis". IEB 2022; Trieste, Italia (Poster presentation).

- Pecori A, Luppieri V, Spedicati B, Santin A, Schito R, Cadenaro M, Girotto G, Maria Pina Concas. "Genetic factors involved in bruxism: the first Genome-Wide Association Study (GWAS) in isolated populations from North-Eastern Italy". ESHG 2023; Glasgow, UK (Poster presentation).
- Santin A, Lenarduzzi S, Nardone GG, Spedicati B, Morgan A, Persichillo M, De Curtis A, Costanzo S, Gialluisi A, Iacoviello L, Gasparini P, Concas MP, Girotto G. "Puzzling out the genetic landscape of Hearing Function (HF): a combined approach of Genome-Wide Association Studies (GWAS) and Transcriptome-Wide Association Studies (TWAS)". ESHG 2023; Glasgow, UK (Poster presentation).
- Spedicati B, Santin A, Nardone GG, Lenarduzzi S, Rubinato E, Graziano C, Garavelli L, Miccoli S, Bigoni S, Morgan A, Girotto G. "Unveiling the genetic bases of Hereditary Hearing Loss (HHL): the application of a multistep diagnostic approach in a large Italian cohort". ESHG 2023; Glasgow, UK (Poster presentation).
- Nardone GG, Santin A, Morgan A, Spedicati B, Concas MP, Girotto G. "Whole Genome Sequencing (WGS) for the molecular diagnosis of Hereditary Hearing Loss (HHL): the underestimated role of Structural Variants (SVs)". ESHG 2023; Glasgow, UK (Poster presentation).
- Girotto G, Spedicati B, Alessandrini B, Della Monica M, Zampino G, Scarano G, Gasparini P, Memo L."Genetic syndromes hiding behind art masterpieces: an intriguing relationship between medicine and arts". ESHG 2023; Glasgow, UK (Poster presentation).
- Tesolin P, Bartoccioni P, Pianigiani G, Morgan A, Caballero M, Ramos I, Gasparini P, Nunes V, Girotto G, Palacin M. "Repurposing drugs to treat SLC7A8 age-related hearing loss". ESHG 2023; Glasgow, UK (Poster presentation).
- Sbaffi C, Spedicati B, Rubinato E, Faletra F, Pellicione G, Zampieri S, Morgan A, Girotto G. "Rapid whole exome sequencing (rWES) in neonatal care in an Italian maternal-children hospital". ESHG 2023; Glasgow, UK (Poster presentation).
- Santin A, Lenarduzzi S, Nardone GG, Spedicati B, Morgan A, Persichillo M, De Curtis A, Costanzo S, Gialluisi A, Iacoviello L, Gasparini P, Concas MP, Girotto G. "Hearing Function: a combined Approach of Genome-Wide Association Studies (TWAS) analysis". IEB Meeting 2023; Londra, UK (Poster presentation).
- Spedicati B, Santin A, Nardone GG, Lenarduzzi S, Paccagnella E, Rubinato E, Morgan A, Girotto G. "Unravelling the Genetic Bases of Hearing Loss: a Multistep and Integrative Approach in a Deeply Characterised Italian Cohort." IEB Meeting 2023; Londra, UK (Poster presentation).
- Pianigiani G, Morgan A, Abidi A, Alonso Jimenez S, Generali M, Spedicati B, Gasparini P, Girotto G, Roccio M. "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". IEB Meeting 2023; Londra, UK (Podium presentation).
- Tesolin P, Bartoccioni P, Pianigiani G, Morgan A, Caballero M, Rojas G, Ramos I, Aloy P, Gasparini P, Nunes Martinez V, Girotto G, Palacin M. "High throughput screening to identify SLC7A8 transcriptional regulators for the treatment of Age-realted hearing loss". IEB Meeting 2023; Londra, UK (Poster presentation).
- Sanna, S, Lenarduzzi S, Busonero F, et al. including Girotto G. "The Women4Health cohort: a multi-omics approach to understand the role of microbiome in women's metabolism". EMBL Symposium 2023; Heidelberg, Germany.
- Pecori A, Zampieri S, Luppieri V, Spedicati B., Santin A, Cadenaro M, Girotto G, Concas MP. "Exploring new genes and risk factors factors associated with bruxism in Italian samples". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Morgan A, Spedicati B, Santin A, Nardone GG, Lenarduzzi S, Paccagnella E, Rubinato E, Girotto G. **"**Unravelling the genetic bases of Hearing Loss (HL): deep phenotyping and high throughput sequencing technologies in an Italian Cohort of patients". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Santin A, Spedicati B, Morgan A, Lenarduzzi S, Tesolin P, Nardone GG, Camarda S, Stevens H, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Concas MP, Zito G, Ricci G, Girotto G. "Endometriosis (EM): still an unsolved genetic dilemma? Whole-Exome Sequencing (WES) analysis and gene discovery

in a highly characterized cohort". SIGU meeting 2023; Rimini, Italia (Poster presentation).

- Santin A, Lenarduzzi S, Nardone GG, Spedicati B, Morgan A, Persichillo MR, De Curtis A, Costanzo S, Gialluisi A, Iacoviello L, Gasparini P, Concas MP, Girotto G. "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". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Spedicati B, Lenarduzzi S, Dal Ferro M, Paldino A, Mazzà M, Zecchin M, D'Errico S, Sinagra G, Gasparini P, Girotto G. "Empowering the application of the molecular autopsy in Sudden Cardiac Deaths (SCD): the experience of the Friuli-Venezia Giulia (FVG) Regional Register". SIGU meeting 2023; Rimini, Italia (Podium presentation).
- Spedicati B, Paccagnella E, Rubinato E, Salvador A, Pelliccione G, Zampieri S, Morgan A, Girotto G. "Rapid Whole Exome Sequencing (rWES) analysis in critically-ill newborns: when a precise and early diagnosis is fundamental for prompt and tailored management". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Nardone GG, Spedicati B, Pecori A, Santin A, Morgan A, Concas MP, Gasparini P, Boscolo-Rizzo P, Girotto G. "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". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Nardone GG, Andrioletti V, Santin A, Morgan A, Spedicati B, Concas MP, Limongelli I, Girotto G. "Structural Variation in short-read Whole Genome Sequencing (srWGS): towards a more accurate detection for clinical practice implementation". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Tesolin P, Spedicati B, Feresin A, Della Monica M, Zampino G, Scarano G, Gasparini P, Memo L, Girotto G. "The new era of arts and genetics: a microscopic study of macroscopic representations". SIGU meeting 2023; Rimini, Italia (Podium presentation).
- Tesolin P, Bartoccioni P, Pianigiani G, Morgan A, Caballero M, Granado GR, Ramos I, Aloy P, Gasparini P, Martinez VN, Girotto G, Palacin M. "Highlighting SLC7A8 Transcriptional Regulators for the Treatment of Age-Related Hearing Loss". SIGU meeting 2023; Rimini, Italia (Podium presentation).
- Zampieri S, Lenarduzzi S, Paldino A, Spedicati B, Mazzà D, Dal Ferro M, Sinagra G, Gasparini P, Girotto G. "Integrated approach for the molecular characterization of 440 Italian patients affected by different forms of hereditary cardiovascular diseases". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Mazzà D, Lenarduzzi S, Morgan A, Zampieri S, Spedicati B, Concas MP, Paccagnella E, Feresin A, Bonati MT, Rubinato E, Dal Ferro M, Paldino A, D'Errico S, Sinagra G, Gasparini P, Girotto G. "Incidentalomi in geni associati a rischio oncologico in pazienti candidati ad esoma per malattie cardiologiche". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Paccagnella E, Feresin A, Bonati MT, Morgan A, Zampieri S, Spedicati B , Bruno I, Carrozzi M , Zanus C , Girotto G, Musante L. "Expanding the molecular and clinical phenotype of the PIG-family disorders". SIGU meeting 2023; Rimini, Italia (Poster presentation).
- Santin A, Spedicati B, Morgan A, Lenarduzzi S, Tesolin P, Nardone GG, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Concas MP, Zito G, Ricci G, Girotto G. "Unravelling the genetic enigma of Endometriosis: novel insights and gene discovery on a deeply characterized cohort". ASHG 2023; Washington, USA.
- Spedicati B, Lenarduzzi S, Dal Ferro M, Paldino A, Mazzà D, D'Errico S, Sinagra G, Gasparini P, Girotto G. "Puzzling out the genetic bases of hereditary cardiovascular diseases: application of an integrative approach in a deeply clinically characterised Italian cohort." ASHG 2023; Washington, USA.
- Nardone GG, Spedicati B, Pecori A, Santin A, Morgan A, Concas MP, Tirelli G, Gasparini P, Boscolo-Rizzo P, Girotto G. "Whole Genome Sequencing in COVID-19 persistent chemosensory dysfunction: a deep characterization of a highly selected Italian cohort". ASHG 2023; Washington, USA.
- Pecori A, Luppieri V, Santin A, Spedicati B, Zampieri S, Cadenaro M, Girotto G,

Concas M.P. “Investigation of environmental and genetic factors associated with bruxism in five genetically isolated populations from North-Eastern Italy”. HGM 2024, Rome, Italy.

- Santin A, Pecori A, Spedicati B, Morgan A, Lenarduzzi S, Tesolin P, Nardone G.G, Camarda S, Stevens H, Pianigiani G, Rosso L.E, Vinerbi E, Bon C, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Campisciano G, Zito G, Sanna S, Concas M.P, Ricci G, Girotto G. **“**Connecting the dots of Endometriosis disease: the role of genetics, diet, and microbiome”. HGM 2024, Rome, Italy.
- Spedicati B, Memo L, Morgan A, Zampieri S, Paccagnella E, Zucca G, Troian M, Agolini E, Dotta A, Rosina E, Pezzani L, Novelli A, Iascone M, Girotto G. “When time matters: application of high-throughput sequencing technologies in critically-ill infants admitted to Neonatal and Pediatric Intensive Care Units (NICU/PICU)”. HGM 2024, Rome, Italy.
- Pianigiani G, Morgan A, Roccio M, Girotto G. “Assessing the functional role of novel gene mutations associated to hearing loss with human iPSC-derived Inner Ear Organoids”. HGM 2024, Rome, Italy.
- Nardone G.G, Andrioletti V, Santin A, Morgan A, Spedicati B, Concas M.P, Limongelli I, Girotto G. “Optimizing Structural Variant Calling: towards a robust and reliable detection from Whole Genome Sequencing (WGS)”. HGM 2024, Rome, Italy.
- Tesolin P, Bartoccioni P, Pianigiani G, Caballero M, Morgan A, Bon C, Granado G.R, Ramos I, Aloy P, Gasparini P, Nunes V, Girotto G, Palacin M. “*SLC7A8* transcriptional regulators identification for the treatment of Age-Related Hearing Loss (ARHL)”. HGM 2024, Rome, Italy.
- Cappellani S, d’Adamo A.P, Spedicati B, Paccagnella E, Zampieri S, Favretto D, Turchetto G, De Santo D, Maso G, Ulivi S, Girotto G, Gasparini P. “The value of SNP-arrays analysis in pregnancy losses: a retrospective study”. HGM 2024, Rome, Italy.
- Girotto G, Zampieri S, Paldino A, Spedicati B, Mazzà D, Turchetto G, Dal Ferro M, Sinagra G, Lenarduzzi S. “Integrated approach for the molecular characterization of 486 Italian patients affected by different forms of hereditary cardiovascular diseases”. HGM 2024, Rome, Italy.
- Zampieri S, Feresin A, Morgan A, Paccagnella E, Rubinato E, Mazzà D, Spedicati B, Stampalija T, Pinamonti M, Bussani R, Gasparini P, Murru F, Girotto G. “The impact of Whole Exome Sequencing for the molecular diagnosis of fetal malformations: the experience of a referral Italian hospital”. HGM 2024, Rome, Italy.
- Santin A, Pecori A, Spedicati B, Morgan A, Lenarduzzi S, Tesolin P, Nardone G.G, Camarda S, Stevens H, Pianigiani G, Rosso L.E, Vinerbi E, Bon C, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Campisciano G, Zito G, Sanna S, Concas M.P, Ricci G, Girotto G. “The molecular riddle of Endometriosis (EM) disorder: filling in the blanks between genetics, diet, and microbiome”. ESHG 2024, Berlin, Germany.
- Spedicati B, Morgan A, Lenarduzzi S, Rubinato E, Zucca G, Troian M, Marangoni D, Girotto G. **“**The diagnostic chameleon of Hereditary Hearing Loss (HHL): a genetic investigation on Non-syndromic Mimics (NSMs)”. ESHG 2024, Berlin, Germany.
- Pianigiani G, Morgan A, Troian M, Turchetto G, Spedicati B, Roccio M, Girotto G. “Modeling genetic forms of hearing loss with Inner Ear Organoids”. ESHG 2024, Berlin, Germany.
- Tesolin P, Bartoccioni P, Pianigiani G, Caballero M, Morgan A, Bon C, Granado G.R, Ramos I, Aloy P, Gasparini P, Nunes V, Girotto G, Palacin M. “High throughput screening: identification of candidate molecules for the treatment of *SLC7A8*- dependent age-related hearing loss (ARHL)”. ESHG 2024, Berlin, Germany.
- Girotto G, Zampieri S, Paldino A, Spedicati B, Turchetto G, Mazzà D, Dal Ferro M, Sinagra G, Lenarduzzi S. “Molecular basis of hereditary cardiovascular diseases (hCVDs) in a cohort of 486 Italian patients: a multi-step approach”. ESHG 2024, Berlin, Germany.
- Liuni R, Bernardinelli E, Huber F, Roesch S, Girotto G, Dossena S. “Novel genetic variants lead to sensorineural hearing loss and Enlarged Vestibular Aqueduct (EVA)”. PMU Science Get Together 2024; Nuremberg, Germany.

- Lenarduzzi S, Morgan A, Troian M, Zampieri S, Nardone G.G, Bon C, Spedicati B, Gasparini P, Girotto G. “Clinical application of Nanopore sequencing in the molecular diagnosis of genetic disorders” SIGU 2024, Padua, Italy.
- Santin A, Pecori A, Spedicati B, Morgan A, Lenarduzzi S, Tesolin P, Nardone G.G, Camarda S, Stevens H, Pianigiani G, Rosso L.E, Vinerbi E, Bon C, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Campisciano G, Zito G, Sanna S, Concas M.P, Ricci G, Girotto G. “Inside-out Endometriosis disease: a 360°-degree view on clinics, genetics, microbiome and diet”. SIGU Padua, Italy.
- Tesolin P, Nardone G.G, Pecori A, Troian M, Santin A, Rosso L.E, Pianigiani G, Spedicati B, Girotto G. “Whole genome sequencing (WGS) in Italian genetic isolates reveals a new CYP2D6 allele”. SIGU 2024, Padua, Italy.
- Santin A, Lenarduzzi S, Nardone G.G, Pecori A, Spedicati B, Morgan A, Persichillo M, De Curtis A, Costanzo S, Gialluisi A, Iacoviello L, Gasparini P, Concas M.P, Girotto G. “Age-Related Hearing Loss (ARHL) genetics at a corner: a GWAS meta-analysis on Italian cohorts”. SIGU 2024, Padua, Italy.
- Cappellani S, Pasquetti D, Ulivi S, Morgan A, Favretto D, d’Adamo A.P, Zampieri S, Girotto G. “Non-invasive prenatal testing (NIPT) for trisomies 13, 18, 21 and sex chromosome aneuploidies: outcomes and clinical experience from 447 consecutive pregnancies”. SIGU 2024, Padua, Italy.
- Zampieri S, Morgan A, Paccagnella E, Rubinato E, Feresin A, Mazzà D, Perino F, Saramin A, Stampalija T, Pinamonti M, Turchetto G, Bussani R, Murru F.M, Spedicati B, Girotto G. “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”. SIGU 2024, Padua, Italy.
- Frisari S, Morgan A, Bonati M.T, Di Marzio G.M, Zanus C, Girotto G, Musante L. “Functional characterization of novel AP3B2 variants identified after systematic WES reanalysis: an example of effective dynamic workflow to reach a diagnosis”. SIGU 2024, Padua, Italy.
- Rosso L.E, Morgan A, Rubinato E, Spedicati B, Girotto G, Pianigiani G. “Functional Characterization of Splicing Variants in Syndromic and Non-Syndromic Hereditary Hearing Loss (HHL) Using Minigene Splicing Assays”. SIGU 2024, Padua, Italy.
- Nardone G.G, Andrioletti V, Santin A, Morgan A, Spedicati B, Troian M, Concas M.P, Limongelli I, Girotto G. “Structural Variant (SV) calling optimization: towards a robust and reliable detection from short read Whole Genome Sequencing (srWGS) data”. SIGU 2024, Padua, Italy.
- Spedicati B, Memo L, Morgan A, Zampieri S, Paccagnella E, Perino F, Frisari S, Troian M, Agolini E, Dotta A, Rosina E, Lucca C, Pezzoli E, Pezzani L, Novelli A, Iascone M, Girotto G. “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”. SIGU 2024, Padua, Italy.
- Spedicati B, Lenarduzzi S, Morgan A, Pianigiani G, Bon C, Rubinato E, Marangoni D, Girotto G. “Unmasking Non-Syndromic Mimics (NSMs) in Hereditary Hearing Loss (HHL) patients: a genetic and clinical investigation in the Italian population” SIGU 2024, Padua, Italy.
- Luppieri V, Pecori A, Santin A, Spedicati B, Zampieri S, Concas M, Girotto G, Cadenaro M. “Identification Of Three Novel Brain-Expressed Genes Potentially Associated With Bruxism”. IADR 2025, Barcelona, Spain.
- Santin A, Pecori A, Nardone G.G, Spedicati B, Lenarduzzi S, Tesolin P, Morgan A, Camarda S, Stevens H, Fasoli L, Mazzà D, Di Lorenzo G, Romano F, Buonomo F, Mangogna A, Zito G, Concas M.P, Ricci G, Girotto G. " Endometriosis (EM) symptoms and extra-oral taste-associated genes (TGs): not only a matter of taste”. ESHG 2025 Milan, Italy
- Spedicati B, Memo L, Morgan A, Zampieri S, Paccagnella E, Perino F, Frisari S, Troian M, Agolini E, Dotta A, Rosina E, Lucca C, Pezzoli L, Pezzani L, Novelli A, Iascone M, Girotto G. “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)”. ESHG 2025 Milan, Italy
- Nardone G.G, Andrioletti V, Santin A, Morgan A, Spedicati B, Troian M, Concas M.P, Limongelli I, Girotto G. “A Hitchhiker guide to Structural Variant (SVs) Calling: a comprehensive benchmark through short-reads (srWGS) and long-reads

(lrWGS) sequencing technologies". ESHG 2025 Milan, Italy

- Rosso L.E, Morgan E, Rubinato E, Spedicati B, Girotto G, Pianigiani G. "Functional Characterization of Splicing Variants in Syndromic and Non-Syndromic Hereditary Hearing Loss (HHL) Using Minigene Splicing Assays". ESHG 2025 Milan, Italy
- Zampieri S, Tesolin P, Nardone G.G, Pecori A, Concas M.P, Troian M, Santin A, Rosso L.E, Pianigiani G, Spedicati B, Girotto G. "Whole genome sequencing (WGS): a powerful tool to gain novel insight into the CYP2D6 gene". ESHG 2025 Milan, Italy
- Tesolin P, Pasquetti P, Santin A, Spedicati B, Memo L, Girotto G. "Art and Genetics: we are not different, we are unique". ESHG 2025 Milan, Italy
- Bortot L, Spedicati B, Paldino A, Lenarduzzi S, Santin A, Dal Ferro M, Sinagra G, Girotto G. "A Rare Case of Hypertrophic-Restrictive Cardiomyopathy with Extensive Myocardial and Valvular Calcifications: Whole-Exome Sequencing (WES) Analysis of an Uncommon Clinical Phenomenon". ESHG 2025 Milan, Italy
- Gasparini V.R, Spedicati B, Pianigiani G, Santin A, Falcomer I, Mazzà D, Paccagnella E, Pasquetti D, Rubinato E, Granata C, Maria Murru F, Pinamonti M, Bussani R, Fantasia I, Stampalija T, Gasparini P, Zampieri S, Girotto G. "Whole exome sequencing (WES) increases the molecular diagnosis of 106 foetuses with structural anomalies negative at chromosomal microarray analysis". ESHG 2025 Milan, Italy
- Frisari S, Zampieri S, Luppieri V, Concas V, Spedicati B, Santin A, Pecori A, Cadenaro M, Girotto G. "Exploring the genetics of molar incisor hypomineralization (MIH): a molecular study on candidate genes involved in human dental enamel development". ESHG 2025 Milan, Italy
- Perino F, Pianigiani G, Pasquetti D, Paccagnella E, Rubinato E, Zampieri S, Gasparini V.R, Girotto G. "Exploring EDA Gene Splicing Variants: Functional Insights, Clinical Implications, and Therapeutic Opportunities". ESHG 2025 Milan, Italy

Language skills

Languages	Speaking	Writing
English	C2	C2
French	A2	A2
Spanish	A2	A2
Portuguese	A1	A1

09/10/25