CURRICULUM VITAE

INFORMAZIONI PERSONALI

Cognome e Nome	**GIROTTO GIORGIA**
Qualifica	**PROFESSORE ASSOCIATO/ DIRIGENTE BIOLOGO CONVENZIONATO**
Incarico attuale	**PROFESSORE ASSOCIATO/ DIRIGENTE BIOLOGO CONVENZIONATO**
Numero telefonico dell'ufficio	**+39 0403785527**
Fax dell'ufficio	**+ 39 0403785540**
Indirizzo e-mail	**giorgia.girotto@burlo.trieste.it**

Titoli di studio e professionali

2021: **Abilitazione Scientifica Nazionale a Professore di prima fascia** (06/A1 Genetica Medica, BANDO D.D. 2175/2018)
2017: **Abilitazione Scientifica Nazionale a Professore di seconda fascia** (06/A1 Genetica Medica, Bando D.D. 1532/2016)
2017: **Specializzazione in Genetica Medica**, Università Degli Studi di Genova, Italia
2013: Iscrizione all'Ordine Nazionale dei Biologi (AA_067918)
2012: **Dottorato di Ricerca in Genetica Molecolare con menzione di Doctor Europeaus**, Università degli Studi di Trieste, Italia
2012: Esame di Stato per Biologi.
2009: **Laurea Magistrale in Biotecnologie Mediche**, Università degli Studi di Trieste, Italia
2006: **Laurea Triennale in Biotecnologie**, Università degli Studi di Trieste, Italia
2003: **Diploma di Liceo Classico**, (Mestre-Venezia, Italia)

Incarico attuale

Da Settembre 2023: Direttore della SS Laboratorio di Genomica e Bioinformatica presso la SC di Genetica Medica dell'IRCCS materno-infantile Burlo Garofolo.
Da Ottobre 2021: Professore associato in Genetica Medica, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste.
Da Gennaio 2018: Dirigente Biologo convenzionato, SC Genetica Medica, Dipartimento dei Servizi e di Diagnostica Avanzata dell'IRCCS materno-infantile Burlo Garofolo.

Partecipa attivamente a corsi di formazione interna ed esterna multidisciplinari con particolare riferimento a tematiche genomiche e bioinformatiche. È coinvolta in prima persona nell'organizzazione del percorso diagnostico-clinico per la gestione dell'analisi dei dati genetici e genomici dei pazienti afferenti alla SC di Genetica Medica.
I risultati positivi della valutazione dell'attività diagnostica (valutazioni Dirigenti ai sensi dell'art. 59 del CCNL Area Sanità del 19/12/2019) presso la SC di Genetica Medica hanno riportato che:

1) Ha completato gli incarichi affidati dimostrando notevoli competenze supportate da una ricca attività scientifica (si veda sezione relativa ad abstract e pubblicazioni) nonché da finanziamenti nazionali e internazionali vinti in prima persona inerenti a malattie ereditarie e multifattoriali e loro studio tramite analisi genetiche/genomiche.
2) Ha partecipato attivamente a procedure di controllo del laboratorio e della qualità clinica delle prestazioni.
3) Ha implementato e promosso innovazioni tecnologiche (con particolare riferimento al campo della genomica e della bioinformatica) consentendo di a) centralizzare l'analisi genetica di alcune patologie (es. ipoacusie, cardiomiopatie, sindromi malformative, neonati critici) rendendo quindi l'Istituto centro di riferimento regionale e nazionale, b) riducendo i tempi di

analisi e migliorando il percorso diagnostico-molecolare.

4) È membro e partecipa attivamente a gruppi di lavoro della Società Italiana di Genetica Umana (es. GdL scienze omiche e citogenomica, malattie multifattoriali), nonché gruppi nazionali per lo studio di sindromi malformative complesse (Rete IDEA) e gruppi di cardiologia e otorinolaringoiatria.
5) Ha organizzato numerosi convegni nazionali e internazionali nel campo della genetica e della genomica essendo Docente di diversi corsi universitari afferenti al Dipartimento di Scienze Mediche Chirurgiche e della Salute, nonché della Scuola di Dottorato in Scienze della Riproduzione e dello sviluppo e della scuola di Specializzazione in Genetica Medica.

Attività di ricerca:

- Coordinatrice di un gruppo di ricerca sulle seguenti tematiche:
    - Tratti e patologie sensoriali (udito, olfatto, vista e gusto) con l'obiettivo di identificare nuovi geni e varianti associati alla fisiologica funzione sensoriale e relativo decadimento.
    - Tratti e patologie complesse, compresi endometriosi, tratti psichiatrici, tratti e patologie odontostomatologiche e patologie cardiometaboliche.
    - Caratterizzazione clinica e gentica degli "Human Knockout" in popolazioni geneticamente isolate.
- Coordinatrice del Consorzio Internazionale G-EAR, la cui attività ha come principale obietttivo quello di studiare le basi genetiche della funzione uditiva e delle ipoacusie.
- Membro di più di 20 consorzi internazionali volti allo studio di tratti e patologie complesse tramite GWAS, contribuendo dalla fase di campionamento all'analisi dei dati.

Attività diagnostica:

Coordinamento dell'analisi dei dati NGS inerenti a malattie ereditarie quali ipoacusia (forme sindromiche e non sindromiche), cardiomiopatie, e patologie sindromiche malformative. Esegue inoltre consulenze genetiche pre-test per numerose patologie ereditarie, e consulenze genetiche per ipoacusia.

Esperienze professionali precedenti

- 2019-2021: **Ricercatore (RTDB)**, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste. Durante questo periodo ha esteso le sue collaborazioni nazionali ed internazionali e proseguito con le attività precedentemente citate, sia sul fronte della ricerca che della diagnostica.
- 2015-2019: **Ricercatore (RTDA)**, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste. Durante questo periodo ha rafforzato la collaborazione di lunga data con i genetisti e gli audiologi del Medical Corporation-HMC e del Sidra Medical Reseach Hospital, all'interno del progetto intitolato "Genetica della sordità nella popolazione del Qatar". Progetti: "Hearing loss in Qatar: High Throughput technologies to identify the molecular basis of deafness and of age-related hearing impairments"; "Molecular bases of hearing loss in Qatar: the final countdown". Grazie a queste collaborazioni e progetti ha visitato più volte l'HMC e, da agosto 2015 a Novembre 2016, è stata Visiting Scientist al Sidra Medical Hospital.
- 2013-2015: **Assegnista di ricerca**, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste. Progetto principale: identificazione di geni coinvolti nelle ipoacusie (forme congenite e tardive) e coordinamento del Consorzio Internazionale G-EAR. Durante questo periodo è stata due mesi Visiting Scientist presso il Center for Genomic Regulation, CRG (Barcellona, Spagna) e sei mesi come Visiting Scientist presso il Mary Lyon Centre (Harwell, Regno Unito) all'interno di un progetto collaborativo sulla presbiacusia dal titolo "Investigating the genetic bases of age-related hearing loss - Human GWAS to mouse models; Proof of Principle". Ha anche organizzato e participato in una spedizione scientifica pilota per raccogliere DNA e fenotipi sensoriali in un'area remota della Mongolia.
- 2010-2012: **Studente di Dottorato in Genetica Molecolare** presso la Scuola di Dottorato in Scienze della Riproduzione e dello Sviluppo, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste. Durante il secondo anno di dottorato ha trascorso sei mesi come Visiting Scientist presso il

Welcome Trust Sanger Insitute (Cambridge-UK) nel "Genetics of deafness laboratory". Inoltre, in questo periodo ha partecipato alla spedizione scientifica "Marco Polo" finalizzata alla raccolta di campioni e all'esecuzione di test audiometrici lungo la Via della Seta.

- 2009**: Incarico di Collaborazione**, presso la SC Genetica Medica dell'IRCCS materno-infantile Burlo Garofolo.
- 2009: **Visiting scientist** presso il National Institute for Medical Research, Londra (Regno Unito), nel laboratorio di Struttura Molecolare.

Attività di insegnamento

Attività di insegnamento

Insegna Genetica Medica nei seguenti corsi di laurea:

- "Medicina e Chirurgia" dal 2020, Università degli Studi di Trieste.
- "Odontoiatria e protesi dentaria" dal 2022, Università degli Studi di Trieste.
- "Logopedia" dal 2022, Università degli Studi di Trieste.
- "Biotecnologie Mediche" dal 2022, Università degli Studi di Trieste.
- "Assistenza Sanitaria" dal 2022 al 2023, Università degli Studi di Trieste.
- "Tecnica della riabilitazione psichiatrica" dal 2016 al 2023, Università degli Studi di Trieste.
- "Psicologia" dal 2018 al 2019, Università degli Studi di Trieste.
- "Igiene Dentale" dal 2019 al 2023, Università degli Studi di Trieste.

Insegna Terapie Innovative in Genetica Medica nei seguenti corsi di laurea:

- Medicina e Chirurgia" dal 2020, Università degli Studi di Trieste.

Insegna inoltre nei seguenti corsi:

- Corso di Dottorato in "Personalised Medicine and Innovative Therapies" dal 2018, Università degli Studi di Trieste.
- Scuola di Specializzazione in "Genetica Medica" (SSM18 and SSM22) dal 2016, Università degli Studi di Trieste.
- Scuola di Specializzazione in "Ginecologia e Ostetricia" (SSM06) dal 2019 al 2021, Università degli Studi di Trieste.
- Scuola di Specializzazione in "Endocrinologia and Malattie del Metabolismo" (SSM25) dal 2022, Università degli Studi di Trieste.
- Master di secondo livello in "Implantologia protesica uditiva" dal 2016 al 2021, Università di Roma "Sapienza".
- Master in "Chirurgia oncoplastica della mammella" dal 2021, Università degli Studi di Trieste.
- Master in "Basic science in Audiology" dal 2021 al XXX, Università degli Studi di Torino e Padova.
- Master in "Scienze uditive dell'età evolutive" nel 2021, Università degli Studi di Perugia.
- Master in "Basic science in audiologia e foniatria" nel 2022, Centro ricerche e studi Amplifon.

Relatrice e correlatrice di tesi:

È stata relatrice e correlatrice delle seguenti tesi di laurea triennale, magistrale, a ciclo unico, di specializzazione e dottorato:

- 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".

Altri incarichi in ambito accademico:

- Membro del Consiglio Didattico del Corso di Dottorato in "Personalised Medicine and Innovative Therapies" dal 2018, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste. Organizzatrice della "PhD Week" dal 2018.
- Membro del Consiglio Didattico della Scuola di Specializzazione in "Genetica Medica" (SSM18 and SSM22), dal 2018, Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste.
- Membro della Commissione di discussione della tesi di laurea in diverse sessioni, nei seguenti corsi di laurea: Medicina, Biologia, Biotecnologie, Biotecnologie Mediche e Genomica Funzionale.
- Delegato per l'area Linguistica dal 2017 al 2021 presso il Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste.
- Delegato per la Commissione Ricerca dal 2021 presso il Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste.

Altri incarichi

- Organizzatrice della spedizione scientifica "Geni Friulani nel Mondo, Brasile 2020"
- Organizzatrice del "Symposium & 57th Inner Ear Biology Workshop – IEB 2022" (10-13/09/2022, Trieste, Italia).
- Componente del Comitato Scientifico della Onlus "Cinzia Vitale" dal 2022
- Membro dei seguenti gruppi di lavoro della Società Italiana di Genetica Umanna (SIGU)
    - "Scienze omiche e citogenomica, genetica prenatale e riproduttiva".
    - "Malattie Multifattoriali".
- Guest Editor per il numero speciale della rivista scientifica "Genes" dal titolo: "Genetics and Epigenetics of Hearing Loss", from 2020 to 2021.
- Guest Editor per il numero speciale della rivista scientifica "Audiology Research" dal titolo: "Genetics of Hearing Loss—Volume II", from 2023.
- Membro del Comitato Scientifico di HUGO meeting 2024

Finanziamenti

- 2015: Finanziamento Scientific Independence of Young Researchers (SIR) (RBSI14AG8P). Titolo del progetto: "Age-related hearing loss: from gene identification to a better molecular diagnosis and prevention" (Ruolo: PI).
- 2016: Finanziamento Junior Scientists Research Experience Program (JSREP07-013-3-006). Titolo del progetto: "Age-related hearing loss in Qatar: a genomic approach to identify causative genes" (Ruolo: PI).
- 2019: Fondazione Beneficentia Stiftung. Titolo del progetto: "Hereditary Hearing Loss: new genes discovery for a preventive strategy and the development of new therapeutic targets" (Ruolo: PI).
- 2019: Progetto MIUR "SENSAGING project "Sensory decays and ageing" (2019-2022) (Ruolo: partecipante).
- 2020: Finanziamento dal titolo "Hearing loss: from genes identification to personalized diagnosis and treatments" vinto presso il Dipartimento di Scienze Mediche, Chirurgiche e della Salute, Università degli Studi di Trieste (Ruolo: PI).
- 2020: Progetto ENDO-2020-23670288 dal titolo "Endometrosis: Pathogenesis of endometriosis: the role of genes, inflammation and environment" presso l'IRCCS Burlo Garofolo, Trieste, Italia (Ruolo: PI).
- 2021: D70-FRA-2021 (Università degli Studi di Trieste).
- 2021: Progetto RC-02-2021 intitolato "A unique cohort to study mechanisms underlying sex differences in cardiometabolic diseases", presso l'IRCCS Burlo Garofolo, Trieste, Italia (Ruolo: PI).
- 2021: Progetto RF-2021-12374963 intitolato "Less genes more genomes: modeling the implementation of integrative -OMICs as a first line tool in the clinical practice" presso l'Ospedale Pediatrico "Bambino Gesù", Roma, Italia (Ruolo: partecipante).
- 2022: Progetto PNRR-MR1-2022-12376811 intitolato "Implementation of a standardized workflow for a more effective management and care of patients with syndromic and isolated intellectual disability" presso l'Ospedale Pediatrico "Bambino Gesù", Roma, Italia (Ruolo: partecipante).

Collaborazioni nazionali ed internazionali

- Partecipazione al consorzio INGI sulle popolazioni geneticamente isolate.
- Collaborazioni con centri di ricerca internazionali per la generazione di modelli animali per studi funzionali sulle perdite uditive (King's college Londra, UK; MRC-Harwell, UK; Harvard Medical School-Howard Hughes Medical Istitute, Dipartimento di Neurobiologia, Boston, USA; Harvard Medical School and Brigham and Women's Hospital, Dipartimento di Patologia, Boston, USA).
- Collaborazione internazionale per lo studio delle perdite uditive della popolazione qatarina (Audiology and Balance Unit, and Molecular Genetics Laboratory and Laboratory of Medicine and Pathology, Hamad Medical Corporation, Doha, Qatar).
- Collaborazioni nazionali con colleghi afferenti a: Università di Milano, Fondazione IRCCS Cà Granda di Milano, Ospedale Maggiore Policlinico di Milano, Università "Cattolica" di Roma, Università degli Studi di Napoli, Università degli Studi di Ferrara, Genetica Medica di Cesena, Genetica Medica di Monza, Università di Bari, Ospedale S.Orsola-Malpighi di Bologna, Università di Ferrara, Università di Padova, IRCCS De Bellis di Castellana Grotte, Università dell'Insubria, Università di Verona, Università di Bologna, CNR, Ospedale di Cagliari.
- Partecipazione a Consorzi internazionali, quali: ICBP (International Consortium of Blood Pressure), CHARGE (Cohorts for Heart and Ageing Research in Genome Epidemiology), GIANT (The Genetic Investigation of Anthropometric Traits), The Visigen Consortium, AlcGen Consortium, CDKGen Consortium, The Reprogen consortium, CRP study, ALSPAC Consortium, CalciGen Consortium, Human Reproductive Behaviour Consortium, NeuroCHARGE Consortium, Dr.Sudha Seshadri (UT Health San Antonio), Dr. Galit Weinstein (Tel Aviv University).
- Collaborazione con il Professor Manuel Palacin (Institute for Research in Biomedicine, Barcelona, Spain) per l'identificazione di molecole in grado di promuovere l'espressione di un gene associato a sordità.
- Partecipazione alla stesura delle line guida nazionale sugli impianti cocleari.

Partecipazione a convegni, seminari e congressi

- Partecipazione a più di otto corsi nazionali ed internazionali dal 2010, inerenti ai temi della genomica per lo studio delle malattie monogeniche e multifattoriali e il campo della statistica e bioinformatica;
- Partecipazione a più di 85 convegni e workshop nazionali ed internazionali dal 2009, inerenti ai temi della genetica medica, della biologia molecolare, della gestione e analisi dei dati di sequenziamento e dello sviluppo e l'implementazione di tecnologie di nuova generazione per l'indagine delle cause molecolari delle malattie genetiche. Nello specifico, sono stati presentati più di 30 lavori come primo o ultimo autore.
- Partecipazione a più di 16 convegni e workshop nazionali ed internazionali dal 2009 come relatore invitato o moderatore di sessione.
- Partecipazione a più di 11 seminari e sei corsi di master come relatore invitato.

Premi e riconoscimenti

Come primo autore

- Premio per il miglior poster presso l'European Meeting Conference 2011 (Amsterdam, The Netherlands, 2011) per il poster dal titolo: "New genes for normal hearing function and age-related hearing loss by genome-wide association and expression studies".
- Premio per la migliore comunicazione orale in genetica clinica, in memoria di Claudio Castellan presso il congresso ESHG/SIGU 2014 (Milano, Italia). Titolo della presentazione: "New Hereditary hearing loss (HHL) genes/mutations identified by high throughput sequencing and genotyping in the Italian and Qatari populations".

Come co-autore o PI del progetto

- Primo premio nella categoria "Biomedicina" per la miglior presentazione orale presso il Qatar Foundation's first Annual Research Forum 2010. Titolo della presentazione: "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 come co-autore della presentazione di Alkowari).
- Premio Spoendlin Junior Award presso il convegno Inner Ear Biology 2015 (G. Girotto come PI del lavoro presentato da Anna Morgan). Titolo della presentazione: "A new targeted re-sequencing panel for unveiling the the genetic causes of age-related hearing loss (ARHL).
- Premio per il miglior poster presso l'European Meeting Conference 2016 (G. Girotto come PI del lavoro presentato da Vuckovic D.). Titolo del poster: "Unravelling human complex traits: the case of hearing function and age related hearing loss".
- Premio per il miglior poster presso il congresso Molecular Biology of Hearing and Deafness (MBHD) 2016 (G. Girotto come PI del lavoro presentato da Anna Morgan). Titolo della presentazione: "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)".
- Premio "Travel Award" presso l'ARO 39th Annual MidWinter Meeting (G. Girotto come PI del lavoro presentato da Anna Morgan). Titolo della presentazione: "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"
- Premio SIGU 2020 per la miglior comunicazione orale (G. Girotto come PI del lavoro presentato da Anna Morgan). Titolo della presentazione: " 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)".
- Premio per il miglior contributo scientifico in genetica clinica, in memoria di Claudio Castellan, al congresso SIGU 2021 (G. Girotto come PI del lavoro presentato da Anna Morgan). Titolo della presentazione: "Clinical application of Whole Exome Sequencing (WES): identification of dual molecular diagnoses in patients affected by Hearing Loss (HL)".
- Premio per il miglior contributo sulle malattie complesse, in memoria di Giuseppe Pilia, al congresso SIGU 2021 (G. Girotto come PI del lavoro presentato da Maria Pina Concas). Titolo della presentazione: "Normal Hearing Function (NHF) and Age-Related Hearing Loss (ARHL): new candidate genes identification from

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

- Premio per il miglior contributo sulle malattie complesse, in memoria di Giuseppe Pilia, al congresso SIGU 2022 (G. Girotto come PI del lavoro presentato da Aurora Santin). Titolo della presentazione: ""Normal Hearing Function (NHF) and Age-Related Hearing Loss (ARHL): new candidate genes identification from Genome-Wide Association Studies (GWAS) in Moli-sani cohort".
- Spoendlin Junior Award 2022 al congresso Inner Ear Biology (G. Girotto come PI del lavoro presentato da Beatrice Spedicati). Titolo della presentazione:" Dual molecular diagnosis in complex hearing loss patients: when a single gene is not enough".
- Premio per il miglior poster al congresso ESHG 2023 (G. Girotto come PI del lavoro presentato da Paola Tesolin). Titolo del poster:" Repurposing drugs to treat *SLC7A8* age-related hearing loss".

Stesura di capitoli in libri

- Consulente scientifico per i libri di testo "Futuri scienziati" e "Accademia delle scienze" (Flaccavento-Romano, Fabbri Editori 2012).
- Consulente scientifico per i libri di testo "Futuri scienziati" e "Accademia delle scienze" (Flaccavento-Romano, Fabbri Editori 2013)
- Capitolo dal titolo "Genetics of Hearing Loss (from Congenital Forms to Presbycusis)" Dipresa S, Fabretto A, Girotto G, Zadro C, Gasparini P, nel libro Dupont, JP, editor. Hearing loss: Classification, Causes and Treatment. Hauppauge, New York: Nova Science Publishers; 2011.
- Capitolo del libro: Ambrosetti U, Castorina P, Gasparini P, Girotto G. "Sordità da cause genetiche". Pubblicato da editore Ambrosetti U. in "Audiologia protesica". Torino, Italia: Edizioni Minerva Medica; 2018.
- Capitolo del libro: Morgan A, Gasparini P, Girotto G. "Hearing loss". Pubbblicato da Lázaro C, Lerner-Ellis J, Spurdle, A. in "Clinical DNA Variant Interpretation: Theory and Practice" a book in the series "Translational and Applied Genomics". Amsterdam: Elsevier/Academic Press; 2021.
- Capitolo del libro: "Guida per cervelli affamati. Perché da bambini odiamo le verdure e altri misteri neurogastronomici che ci rendono umani". Pubblicato da C, Rossi SE in "Il Saggiatore"; 2021.

Attività di divulgazione

Rilascio di numerose interviste per quotidiani locali e nazionali (ad es. Il Piccolo, 24oreNews.it, ilFriuli.it, triestecafe.it), radio, canali televisivi (ad es. Oggi Scienza TV, Rai 3 Scienza, La Repubblica, Il Giornale dei Biologi, Il Bo Live UniPD, Mercurio Associazione Economica Italo-Tedesca)

- 2021: "Trieste Next: spazio 13. Endometriosi, genetica e ambiente. Nuovi scenari per la diagnosi e la cura" (Trieste, Italia).
- 2021: Relatrice all'evento Premio Papa Enrnest Hemingway durante la sessione "L'Approfondimento" (Caorle, Italia).
- 2022: Intervista per il giornale "La Stampa - Tutto Scienze". Titolo: "Perché la tentazione del genoisimo è un'illusione pericolosa?".
- 2022: Intervista per la rubrica "Scienza" del Piccolo di Trieste.
- 2022: Relatrice presso il "Trieste Science+Fiction Festival". Titolo della presentazione: "Eugenetica e genoismo: fiction o realtà? "(Trieste, Italia).

Inoltre, ha partecipato attivamente ad eventi pubblici, fra cui:

- Eventi sociali locali
- Eventi sportivi sponsorizzati.
- Eventi di beneficenza e raccolta fondi.
- Festival della scienza.

Pubblicazioni

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.
2. Pecci A, Panza E, De Rocco D, Pujol-Moix N, Girotto G, Podda L, Paparo C, Bozzi V, Pastore A, Balduini CL, Seri M, Savoia A. "MYH9 related disease: four novel mutations of the tail domain of myosin-9 correlating with a mild clinical phenotype." Eur J Haematol. 2010 Apr; 84(4):291-7. doi: 10.1111/j.1600-0609.2009.01398.x, PMID: 20002731.

3. Girotto G, Pirastu N, Sorice R, Biino G, Campbell H, d'Adamo AP, Hastie ND, Nutile T, Polasek O, Portas L, Rudan I, Ulivi S, Zemunik T, Wright AF, Ciullo M, Hayward C, Pirastu M, Gasparini P. "Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways". J Med Genet. 2011 Jun;48(6):369-74. doi: 10.1136/jmg.2010.088310, PMID: 21493956.
4. Girotto G, Pirastu N, Gasparini A, d'Adamo AP, Gasparini P. "Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road". Audiological Medicine. December 2011; 9(4): 135–140.
5. Khalifa Alkowari M, Girotto G, Abdulhadi K, Dipresa S, Siam R, Najjar N, Badii R, Gasparini P. "GJB2 and GJB6 genes and the A1555G mitochondrial mutation are only minor causes of nonsyndromic hearing loss in the Qatari population". Int J Audiol. 2012 Mar;51(3):181-5. doi: 10.3109/14992027.2011.625983, PMID: 22103400.
6. McQuillan R, Eklund N, Pirastu N et al. (including Girotto G). "Evidence of Inbreeding Depression on Human Height". PLoS Genet. 2012;8(7):e1002655. DOI: 10.1371/journal.pgen.1002655, PMID: 22829771.
7. Boraska V, Jeroncic A, Colonna V, et al. (including Girotto G). "Genome-wide meta-analysis of common variant differences between men and women". Hum Mol Genet. 2012 Nov 1; 21(21):4805-15. doi: 10.1093/hmg/dds304, PMID: 22843499.
8. van der Harst P, Zhang W, Mateo Leach I et al. (including Girotto G). "Seventy-five genetic loci influencing the human red blood cell". Nature. 2012 Dec 20; 492(7429):369-75. doi: 10.1038/nature11677, PMID: 23222517.
9. Nolan LS, Maier H, Hermans-Borgmeyer I, Girotto G, Ecob R, Pirastu N, Cadge BA, Hübner C, Gasparini P, Strachan DP, Davis A, Dawson SJ. "Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice". Neurobiol Aging. 2013; 34(8):2077.e1-9. doi: 10.1016/j.neurobiolaging.2013.02.009, PMID: 23540940.
10. O'Seaghdha CM, Wu H, Yang Q, et al. (including Girotto G.). "Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations". PLoS Genet. 2013; 9(9):e1003796. doi: 10.1371/journal.pgen.1003796, PMID: 24068962.
11. Vuckovic D, Biino G, Panu F, Pirastu M, Gasparini P, Girotto G. "Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee". Hearing, Balance and Communication. 2013; 11: 218–223.
12. Paul DS, Albers CA, Rendon A, Voss K, Stephens J, HaemGen Consortium (Girotto G. being a member of this consortium), van der Harst P, Chambers JC, Soranzo N, Ouwehand WH, Deloukas P. "Maps of open chromatin highlight cell type-restricted patterns of regulatory sequence variation at hematological trait loci". Genome Res. 2013 Jul; 23(7):1130-41. doi: 10.1101/gr.155127.113, PMID: 23570689.
13. Girotto G, Abdulhadi K, Buniello A, Vozzi D, Licastro D, d'Eustacchio A, Vuckovic D, Alkowari MK, Steel KP, Badii R, Gasparini P. "Linkage study and exome sequencing identify a BDP1 mutation associated with Hereditary Hearing Loss". PLoS One. 2013 Dec 2; 8(12):e80323. doi: 10.1371/journal.pone.0080323, PMID: 24312468.
14. Faletra F, Girotto G, D'Adamo AP, Vozzi D, Morgan A, Gasparini P. "A Novel P2RX2 Mutation In An Italian Family Affected By Autosomal Dominant Nonsyndromic Hearing Loss". Gene. 2014 Jan 25; 534(2):236-9. doi: 10.1016/j.gene.2013.10.052, PMID: 24211385.
15. Girotto G, Vuckovic D, Buniello A, Lorente-Cánovas B, Lewis M, Gasparini P, Steel KP. "Expression and replication studies to identify new candidate genes involved in normal hearing function". PLoS One. 2014 Jan 14; 9(1):e85352. doi: 10.1371/journal.pone.0085352, PMID: 24454846.
16. Olden M, Corre T, Hayward C, Toniolo D, Ulivi S, Gasparini P, Pistis G, Hwang SJ, Bergmann S, Campbell H, Cocca M, Gandin I, Girotto G, Glaudemans B, Hastie ND, Loffing J, Polasek O, Rampoldi L, Rudan I, Sala C, Traglia M, Vollenweider P, Vuckovic D, Youhanna S, Weber J, Wright AF, Kutalik Z, Bochud M, Fox CS, Devuyst O. "Common Variants in UMOD Associate with Urinary Uromodulin Levels: A Meta-Analysis". J Am Soc

Nephrol. doi: 10.1681/ASN.2013070781, PMID: 24578125.

17. Vozzi D, Morgan A, Vuckovic D, D'Eustacchio A, Abdulhadi K, Rubinato E, Badii R, Gasparini P, Girotto G. "Hereditary Hearing loss: a 96 genes Targeted Sequencing protocol reveals novel alleles in a series of Italian and Qatari patients". Gene. 2014. doi: 10.1016/j.gene.2014.03.033, PMID: 24657061.
18. Vuckovic D, Biino G, Panu F, Pirastu M, Gasparini P, Girotto G. "Age-related hearing loss and level of education: an epidemiological study on a large cohort of isolated populations". Hearing, Balance and Communication. 2014; 12: 94–98.
19. Girotto G, Mezzavilla M, Abdulhadi K, Vuckovic D, Vozzi D, Khalifa Alkowari M, Gasparini P, Badii R. "Consanguinity and Hereditary Hearing loss in Qatar". Hum Hered. 2014; 77(1-4):175-82. doi: 10.1159/000360475, PMID: 25060281.
20. Wolber LE, Girotto G, Buniello A, Vuckovic D, Pirastu N, Lorente-Cánovas B, Rudan I, Hayward C, Polasek O, Ciullo M, Mangino M, Steves C, Concas MP, Cocca M, Spector TD, Gasparini P, Steel KP, Williams FM. "Salt-inducible kinase 3, SIK3, is a new gene associated with hearing." Hum Mol Genet. 2014 Jul 24. pii: ddu346, doi: 10.1093/hmg/ddu346, PMID: 25060954.
21. Mezzavilla M, Vozzi D, Pirastu N, Girotto G, d'Adamo P, Gasparini P, Colonna V. "Genetic landscape of populations along the Silk Road: admixture and migration patterns". BMC genetics. 2014. doi:10.1186/s12863-014-0131-6, PMID: 25476266.
22. Lenarduzzi S, Vozzi D, Morgan A, Rubinato E, D'Eustacchio A, Osland T, Rossi C, Graziano C, Castorina P, Ambrosetti U, Morgutti M, Girotto G. "Usher Syndrome: an effective sequencing approach to establish a genetic and clinical diagnosis". Hear Res. 2015 Feb; 320:18-23. doi: 10.1016/j.heares.2014.12.006, PMID: 25575603.
23. Zonta F, Girotto G, Buratto D, Crispino G, Morgan A, Abdulhadi K, Alkowari M, Badii R, Gasparini P, Mammano F. "The p.Cys169Tyr variant of connexin 26 is not a polymorphism". Hum Mol Genet. 2015 May 1; 24(9):2641-8. doi: 10.1093/hmg/ddv026, PMID: 25628337.
24. Mezzavilla M, Vozzi D, Badii R, Khalifa Alkowari M, Abdulhadi K, Girotto G, Gasparini P. "Increased Rate of Deleterious Variants in Long Runs of Homozygosity of an Inbred Population from Qatar". Hum Hered. 2015 Feb 21; 79(1):14-19. doi: 10.1159/000371387, PMID: 25720536.
25. Joshi PK, Esku T, Mattsson H, et al. (including Girotto G). "Directional dominance on stature and cognition in diverse human populations". Nature. 2015 Jul 1. doi: 10.1038/nature14618, PMID: 26131930.
26. Vuckovic D, Dawson S, Scheffer D, Rantanen T, Morgan A, Di Stazio M, Vozzi D, Nutile T, Concas MP, Biino G, Nolan, Bahl A, Loukola A, Viljanen A, Davis A, Ciullo M, Corey DP, Pirastu M, Gasparini P, Girotto G. "Genome-wide association analysis on Normal Hearing function identifies PCDH20 and SLC28A3 as candidates for hearing function and loss". Hum. Mol. Genet. 2015. doi: 10.1093/hmg/ddv279, PMID: 26188009.
27. Harrison S, Lewis SJ, Hall AJ, Vuckovic D, Girotto G, Martin RM, Adams JC. "Association of SNPs in LCP1 and CTIF with hearing in 11-year-old children: Findings from the Avon Longitudinal Study of Parents and Children (ALSPAC) birth cohort and the G-EAR consortium". BMC Medical Genomics BMC Med Genomics. 2015 Aug 12; 8:48. doi: 10.1186/s12920-015-0112-2, PMID: 26264041.
28. Day FR, Ruth KS, Thompson DJ et al. (including Girotto G). "Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair". Nat Genet. 2015 Sep 28. doi: 10.1038/ng.3412, PMID: 26414677.
29. Girotto G, Scheffer DI, Morgan A, Vozzi D, Rubinato E, Di Stazio M, Muzzi E, Pensiero S, Giersch AB, Corey DP, Gasparini P. "PSIP1/LEDGF: a new gene involved in sensorineural progressive hearing loss and variable eye phenotypes". Scientific reports Sci Rep. 2015 Dec; 22;5:18568. doi: 10.1038/srep18568, PMID: 26689366.
30. Cocca M, Bedognetti D, La Bianca M, Gasparini P, Girotto G. "Pharmacogenetics driving personalized medicine: analysis of genetic

polymorphisms related to breast cancer medications in Italian isolated populations". J Transl Med. 2016 Jan 22;14(1):22. doi: 10.1186/s12967-016-0778-z, PMID: 26801900.

31. Okbay A, Beauchamp JP, Fontana MA et al. (including Girotto G). "Genome-wide association study identifies 74 loci associated with educational attainment". Nature. 2016 May 26; 533(7604):539-42. doi: 10.1038/nature17671, PMID: 27225129.
32. Barban N, Jansen R, de Vlaming R et al. (including Girotto G). "Genome-wide analysis identifies 12 loci influencing human reproductive behaviour". Nat Genet. 2016 Dec; 48(12):1462-1472. doi: 10.1038/ng.3698, PMID: 27798627.
33. Marioni RE, Ritchie SJ, Joshi PK, et al. (including Girotto G, as a member of the Social Science Genetic Association Consortium). "Genetic variants linked to education predict longevity". Proc Natl Acad Sci U S A. 2016 Nov 22; 113(47):13366-13371. doi: 10.1073/pnas.1605334113, PMID: 27799538.
34. Palombo F, Al-Wardy N, Ruscone GA, Oppo M, Kindi MN, Angius A, Al Lamki K, Girotto G, Giangregorio T, Benelli M, Magi A, Seri M, Gasparini P, Cucca F, Sazzini M, Al Khabori M, Pippucci T, Romeo G. "A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman". J Hum Genet. 2016 Oct 13. doi: 10.1038/jhg.2016.120, PMID: 27734841.
35. Warren HR, Evangelou E, Cabrera CP et al. (including Girotto G). "Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk". Nat Genet. 2017 Jan 30. doi: 10.1038/ng.3768, PMID: 28135244.
36. Marouli E, Graff M, Medina-Gomez C, et al. (including Girotto G). "Rare and low-frequency coding variants alter human adult height". Nature. 2017 Feb 9; 542(7640):186-190. doi: 10.1038/nature21039, PMID: 28146470.
37. Gorski M, van der Most PJ, Teumer A, et al. (including Girotto G). "1000 Genomes-based meta-analysis identifies 10 novel loci for 1 kidney function". Scientific Reports. 2017. doi:10.1038/srep45040, PMID: 28452372.
38. Alkowari M, Vozzi D, Baghat S, Krishnamoothy N, Morgan A, Hayder Y, Logendra B, Najjar N, Gandin I, Gasparini P, Badii R, Girotto G, Abdulhadi K. "Targeted sequencing identifies novel variants involved in Autosomal Recessive Hereditary Hearing Loss in Qatari families". Mutation Research. 2017 Aug. doi: 10.1016/j.mrfmmm.2017.05.001, PMID: 28501645.
39. Wain LV, Vaez A, Jansen R, et al. (including Girotto G). "Novel blood pressure locus and gene discovery using GWAS and expression datasets from blood and the kidney". Hypertension. 2017 Jul 24. pii: HYPERTENSIONAHA.117.09438, doi:10.1161/HYPERTENSIONAHA.117.09438, PMID: 28739976.
40. Espino-Gaurch M, Font-Llitjós M, Murillo-Cuesta S, Errasti-Murugarren E, Celaya AM, Girotto G, Vuckovic D, Mezzavilla M, Vilches C, Bodoy S, Sahún I, González L, Prat E, Zorzano A, Dierssen M, Varela-Nieto I, Gasparini P, Palacín M, Nunes V. "Mutations in L-type amino acid transporter-2 support SLC7A8 as a novel gene involved in Age-Related Hearing Loss". Elife. 2018 Jan 22; 7. pii: e31511, doi: 10.7554/eLife.31511, PMID: 29355479.
41. Hysi P, Valdes AM, Liu F, et al. (including Girotto G). "Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability." Nat Genet. 2018 May; 50(5):652-656. doi: 10.1038/s41588-018-0100-5, PMID: 29662168.
42. Vuckovic D, Mezzavilla M, Cocca M, Morgan A, Brumat M, Catamo E, Concas MP, Biino G, Franzè A, Ambrosetti U, Pirastu M, Gasparini P, Girotto G. "Whole-genome sequencing reveals new insights into Age Related Hearing Loss: cumulative effects, pleiotropy and the role of selection". Eur J Hum Genet. 2018; 26(8):1167-1179. doi: 10.1038/s41431-018-0126-2, PMID: 29725052.
43. Lee JJ, Wedow R, Okbay A, et al., (including Girotto G, as member of the COGENT (Cognitive Genomics Consortium); Social Science Genetic Association Consortium). "Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million

individuals." Nat Genet. 2018 Aug; 50(8):1112-1121. doi: 10.1038/s41588-018-0147-3, PMID: 30038396.

44. Morgan A, Vuckovic D, Krishnamoorthy N, Rubinato E, Ambrosetti U, Castorina P, Franzè A, Vozzi D, La Bianca M, Cappellani S, Di Stazio, M, Gasparini P, Girotto G. "Next-generation sequencing identified SPATC1L as a possible candidate gene for both early-onset and age-related hearing loss". Eur J Hum Genet. 2019; 27(1):70-79. doi: 10.1038/s41431-018-0229-9, PMID: 30177775.
45. Fetoni AR, Zorzi V, Paciello F, Ziraldo G, Raspa M, Scavizzia F, Salvatore AM, Crispino G, Tognola G, Gentile G, Spampinato AG, Cuccaro D, Guarnaccia M, Morello G, Van Camp G, Fransen E, Brumat M, Girotto G, Paludetti G, Gasparini P, Cavallaro S, Mammano F. "Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway". Redox Biol. 2018 Aug 7; 19:301-317. doi: 10.1016/j.redox.2018.08.002, PMID: 30199819.
46. Evangelou E, Warren H, Mosen-Ansorena D, et al. (including Girotto G). "Genetic analysis of over one million people identifies 535 new loci associated with blood pressure traits". Nat Genet. 2018 Sep 17. doi: 10.1038/s41588-018-0205-x, PMID: 30224653.
47. Di Stazio M, Collesi C, Vozzi D, Morgan A, Liu W, Myers M, Rubinato E, Girotto G, D'Adamo P, Giacca M, Gasparini P. "TBL1Y: a new gene involved in syndromic hearing loss". Eur J Hum Genet. 2018 Oct 19. doi: 10.1038/s41431-018-0282-4, PMID: 30341416.
48. Morgan A, Lenarduzzi S, Cappellani S, Pecile V, Morgutti M, Orzan E, Ghiselli S, Ambrosetti U, Brumat M, Gajendrarao P, La Bianca M, Faletra F, Grosso E, Sirchia F, Sensi A, Graziano C, Seri M, Gasparini P, Girotto G. "Genomic studies in a large cohort of hearing impaired Italian patients revealed several new alleles, a rare case of uniparental disomy (UPD) and the importance to search for copy number variations". Front Genet. 2018; 9:681. doi: 10.3389/fgene.2018.00681, PMID: 30622556.
49. Karlsson Linnèr R, Biroli P, Kong E, et al. (including Girotto G). "Genome-wide association analyses of risk tolerance and risky behaviors in over 1 million individuals identify hundreds of loci and shared genetic influences". Nat Genet. 2019 Feb; 51(2):245-257. doi: 10.1038/s41588-018-0309-3, PMID: 30643258.
50. Justice AE, Karaderi T, Highland HM, et al. (including Girotto G). "Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution". Nat Genet. 2019 Feb 18. doi: 10.1038/s41588-018-0334-2, PMID: 30778226.
51. Girotto G, Morgan A, Krishnamoorthy N, Cocca M, Brumat M, Bassani S, La Bianca M, Di Stazio M, Gasparini P. "Next Generation Sequencing and Animal Models Reveal SLC9A3R1 as a New Gene Involved in Human Age-Related Hearing Loss". Front Genet. 2019; 10:142. doi: 10.3389/fgene.2019.00142, PMID: 30863428.
52. Noordam R, Young WJ, Salman R, et al. (including Girotto G). "Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals". J Am Coll Cardiol. 2019 Jun 25;73(24):3118-3131. doi: 10.1016/j.jacc.2019.03.519.
53. Wuttke M, Li Y, Li M, et al. (including Girotto G). "A catalog of genetic loci associated with kidney function from analyses of a million individuals". Nat Genet. 2019 Jun; 51(6):957-972. doi: 10.1038/s41588-019-0407-x.
54. Lenarduzzi S, Morgan A, Faletra F, Cappellani S, Morgutti M, Massimo M, Peruzzi A, Ghiselli S, Ambrosetti U, Graziano C, Seri M, Gasparini P, Girotto G. "Next Generation Sequencing study in a cohort of Italian patients with Syndromic Hearing loss". Hear Res. 2019; 381:107769. doi: 10.1016/j.heares.2019.07.006. Epub 2019 Jul 13.
55. Morgan A, Koboldt DC, Barrie ES, Erin C, Mezzavilla M, Faletra F, Moshe TM, Wilson RK, Manickam K, Gasparini P, Dell'Orco D, Girotto G. "Mutations in PLS1, encoding fimbrin, cause autosomal dominant non-syndromic hearing loss". Hum Mutat 2019; 40(12): 2286-2295. doi: 10.1002/humu.23891.
56. Tin A, Marten J, Halperin-Kuhns, et al. (including Girotto G). "Target genes, variants, tissues and transcriptional pathways influencing human serum urate

levels". Nat Genet. 2019; 51(10):1459-1474. doi: 10.1038/s41588-019-0504-x.

57. Nagtegaal AP, Broer L, Zilhao NR, et al. (including Girotto G). "Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment". Sci Rep. 2019; 9(1):15192. doi: 10.1038/s41598-019-51630-x.
58. Clark DW, Okada Y, Moore K, et al. (including Girotto G). "Associations of autozygosity with a broad range of human phenotypes". Nat Commun. 2019; 10(1):4957. doi: 10.1038/s41467-019-12283-6.
59. Cocca M, Barbieri C, Concas MP, Robino A, Brumat M, Gandin I, Trudu M, Sala CF, Vuckovic D, Girotto G, Matullo G, Polasek O, Kolčić I, Gasparini P, Soranzo N, Toniolo D, Mezzavilla M. "A bird's-eye view of Italian genomic variation through whole-genome sequencing". Eur J Hum Genet. 2019. doi: 10.1038/s41431-019-0551-x.
60. Ingham NJ, Rook V, Di Domenico F, James E, Lewis MA, Girotto G, Buniello A, Steel KP. "Functional analysis of candidate genes from genome-wide association studies of hearing". Hear Res. 2020; 387:107879. doi: 10.1016/j.heares.2019.107879.
61. Morgan A, Pelliccione G, Ambrosetti U, Dell'Orco D, Girotto G. "SLC12A2: a new gene associated with autosomal dominant Non-Syndromic hearing loss in humans". Hearing, Balance and Communication. 2020; 18 (2): 149-151. doi: 10.1080/21695717.2020.1726670.
62. Di Stazio M, Morgan A, Brumat M, Bassani S, Dell'Orco D, Marino V, Garagnani P, Giuliani C, Gasparini P, Girotto G. "New age-related hearing loss candidate genes in humans: an ongoing challenge". Gene. 2020. doi: 10.1016/j.gene.2020.144561.
63. de Las Fuentes L, Sung Y, Noordam R, et al. (including Girotto G). "Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci". Mol Psychiatry. 2021 June; 26: 2111-2125. doi: 10.1038/s41380-020-0719-3.
64. Faletra F, Morgan A, Ghiselli S, Murru FM, Girotto G. "Hearing Loss and Brain Abnormalities due to pathogenic mutations in ADGRV1 gene: a case report". Hearing, Balance and Communication. 2020; 18(3):196-198. doi: 10.1080/21695717.2020.1807255.
65. Piatti G, Ambrosetti U, Robino A, Girotto G, Gasparini P. "Primary Ciliary Dyskinesia: The Impact of Taste Receptor (TAS2R38) Gene Polymorphisms on Disease Outcome and Severity". International Archives of Allergy and Immunology. 2020; 181(9): 727-731. doi: 10.1159/000508938, PMID: 32659773.
66. Morgan A, Gasparini P, Girotto G. "Molecular testing for the study of non-syndromic hearing loss". Hearing, Balance and Communication. 2020; 18(4): 270-277. doi: 10.1080/21695717.2020.1807260.
67. Morgan A., Lenarduzzi S., Spedicati B., Cattaruzzi E., Murru FM, Pelliccione G, Mazzà D, Zollino M, Graziano C, Ambrosetti U, Seri M, Faletra F, Girotto G. "Lights and shadows in the genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population". Genes (Basel). 2020; 11(11):1-16, Art. No. 1237. doi: 10.3390/genes11111237.
68. Mathieson I, Day F, Barban N, et al. (including Girotto G, as a member of the Human Reproductive Behaviour Consortium). "Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus". bioRxiv doi: https://doi.org/10.1101/2020.05.19.104455.
69. Comar M, Brumat M, Concas MP, Argentini G, Bianco A, Bicego L, Bottega R, Carli P, Cassone A, Catamo E, Cocca M, Del Pin M, Di Stazio M, Feresin A, La Bianca M, Morassut S, Morgan A, Pelliccione G, Petix V, Ragusa G, Robino A, Russian S, Spedicati B, Suergiu S, Urriza M, Vascotto F, Toscani P, Girotto G, Gasparini P. "COVID-19 experience: first Italian survey on healthcare staff members from a Mother-Child Research hospital using combined molecular and rapid immunoassays test". bioRxiv doi: https://doi.org/10.1101/2020.04.19.20071563.
70. Ciorba A, Corazzi V, Melegatti M, Morgan A, Pelliccione G, Girotto G, Bigoni S. "Non-syndromic sensorineural Prelingual and Postlingual Hearing

Loss due to COL11A1 gene mutation". Journal of International Advanced Otology. 2021; 17 (1): 81-83. doi: 10.5152/iao.2020.8179.

71. Simcoe M, Valdes A, Liu F, et al. (including Girotto G). "Genome-wide association study in almost 195,000 individuals identifies more than 50 novel genetic loci for eye colour". Science Advances. 2021; 7 (11), art. no. abd1239. doi: 10.1126/sciadv.abd1239.
72. Morgan A, Dipresa S, Turolla L, La Bianca M, Faletra F, Girotto G. "A new case of TAR syndrome confirms the importance of noncoding variants in the etiopathogenesis of the disease". Human Mutation. 2021; 42(2): 213-215. doi: 10.1002/humu.24145.
73. Spedicati B, Cocca M, Palmisano R, Faletra F, Barbieri C, Francescatto M, Mezzavilla M, Morgan A, Pelliccione G, Gasparini P, Girotto G. "Natural Human Knockouts and Mendelian disorders: deep phenotyping in Italian isolates". European Journal of Human Genetics. 2021. doi: 10.1038/s41431-021-00850-9.
74. Concas MP, Cocca M, Francescatto M, Battistuzzi T, Spedicati B, Feresin A, Morgan A, Gasparini P, Girotto G. "The role of knockout Olfactory Receptors Genes in Smell Perception". Genes. 2021; 12(5), 631. doi: https://doi.org/10.3390/genes12050631.
75. Concas MP, Morgan A, Pelliccione G, Gasparini P, Girotto G. "Genetics, odor perception and food liking: the intriguing role of cinnamon". Food Quality and Preference. 2021; 93, 104277. doi: doi.org/10.1016/j.foodqual.2021.104277.
76. Bassani S, Beelen E, Rossel M, Voisin N, Morgan A, Arribat Y, Chatron N, Chrast J, Cocca M, Delprat B, Faletra F, Giannuzzi G, Guex N, Machavoine R, Pradervand S, Smits JJ, van de Kamp JM, Ziegler A, Amati F, Marlin S, Kremer H, Locher H, Maurice T, Gasparini P, Girotto G, Reymond A. "Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss". Human Molecular Genetics. 2021; ddab145. doi: https://doi.org/10.1093/hmg/ddab145.
77. Mills MC, Tropf F, Brazel D, et al. (including Girotto G). "Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour". Nat Hum Behav. 2021; 5:1717-1730. doi: 10.1038/s41562-021-01135-3.
78. Concas MP, Morgan A, Serra F, Nagtegaal AP, Oosterloo BC, Seshadri S, Heard-Costa N, Van Camp G, Fransen E, Francescatto M, Logroscino G, Sardone R, Quaranta N, Gasparini P, Girotto G. "Hearing Function: Identification of New Candidate Genes Further Explaining the Complexity of This Sensory Ability." Genes (Basel). 2021 Aug 10;12(8):1228. doi: 10.3390/genes12081228. PMID: 34440402 Free PMC article.
79. Di Cesare T, Rossi G, Girotto G, Di Nardo W. "Benefit of cochlear implantation in a patient with Myhre syndrome". BMJ Case Rep. 2021 Aug 25;1 4(8):e243164. doi: 10.1136/bcr-2021-243164. PMID: 34433528
80. Tesolin P, Fiorino S, Lenarduzzi S, Rubinato E, Cattaruzzi E, Ammar L, Castro V, Orzan E, Granata C, Dell'Orco D, Morgan A, Girotto G. "Pendred Syndrome, or Not Pendred Syndrome? That Is the Question." Genes (Basel). 2021 Oct 1;12(10):1569. doi: 10.3390/genes12101569.
81. Tesolin P, Morgan A, Notarangelo M, Ortore RP, Concas MP, Notarangelo A, Girotto G. "Non-Syndromic Autosomal Dominant Hearing Loss: The First Italian Family Carrying a Mutation in the NCOA3 Gene". Genes. 2021; 12(7), 1043. doi: https://doi.org/10.3390/ genes12071043.
82. Young W, Lahrouchi N, Isaacs A, et al. (including Girotto G). "Genetic analyses of the QT interval and its components in over 250K individuals identifies new loci and pathways affecting ventricular depolarization and repolarization". 2021. doi: 10.1101/2021.11.04.21265866.
83. Graham SE, Clarke S, Wu K, et al. (including Girotto G). "The power of genetic diversity in genome-wide association studies of lipids". 2021. Nature 600(7890): 675-679. doi: 10.1038/s41586-021-04064-3.
84. Concas MP, Minelli A , Aere S, Morgan A , Tesolin P , Gasparini P, Gennarelli M, Girotto G. "Genetic Dissection of Temperament Personality Traits in Italian Isolates". 2022. Genes 13(1):4. doi.org/

10.3390/genes13010004).

85. Morgan A, Faletra F, Severi G, La Bianca M, Licchetta L, Gasparini P, Graziano C, Girotto G. “There Is More Than Meets the Eye: Identification of Dual Molecular Diagnosis in Patients Affected by Hearing Loss”. 2022. Biomedicines 10(1):12. doi.org/10.3390/biomedicines10010012.
86. Joseph CB, Mariniello M, Yoshifuji A, et al. (including Girotto G). “Meta-GWAS Reveals Novel Genetic Variants Associated with Urinary Excretion of Uromodulin”. J Am Soc Nephrol. 2022; 33(3):511-529. doi: 10.1681/ASN.2021040491, PMID: 35228297.
87. Concas MP, Morgan A, Tesolin P, Santin A, Girotto G, Gasparini P. “Sensory Capacities and Eating Behavior: Intriguing Results from a Large Cohort of Italian Individuals”. Foods. 2022; 11(5). doi: 10.3390/foods11050735.
88. Piatti G, Ambrosetti U, Aldè M, Girotto G, Concas MP, Torretta S. “Chronic Rhinosinusitis: *T2R38* Genotyping and Nasal Cytology in Primary Ciliary Dyskinesia”. Laryngoscope. 2022. doi: 10.1002/lary.30112; PMID: 35312075.
89. Fongang B, Weinstein G, Gudjonsson A, et al. (including Girotto G). “A meta-analysis of genome-wide association studies identifies new genetic loci associated with all-cause and vascular dementia”. Alzheimer's & dementia: the journal of the Alzheimer's Association. 2022; 17(54): e056081. doi: 10.1002/alz.056081.
90. Trpchevska N, Freidin M, Broer L, et al. (including Girotto G). “Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss”. Am J Hum Gen. 2022; S0002-9297(22)00158-6. doi: https://doi.org/10.1016/j.ajhg.2022.04.010, PMID: 35580588.
91. Okbay A, Wu Y, Wang N, et al. (including Girotto G). “Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals”. Nat Gen. 2022; 54(4):437-449. doi: 10.1038/s41588-022-01016-z, PMID: 35361970.
92. Musante L, Faletra F, Meier K, Tomoum H, Najarzadeh Torbati P, Blair E, North S, Gärtner J, Diegmann S, Beiraghi Toosi M, Ashrafzadeh F, Ghayoor Karimiani E, Murphy D, Murru FM, Zanus C, Magnolato A, La Bianca M, Feresin A, Girotto G, Gasparini P, Costa P, Carrozzi M. “TTC5 syndrome: clinical and molecular spectrum of a severe and recognizable condition”. American Journal of Medical Genetics Part A. 2022. doi: 10.1002/ajmg.a.62852, PMID: 35670379.
93. Winkler TW, Rasheed H, Teumer A, et al. (including Girotto G). “Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals”. Commun Biol. 2022 Jun; 13;5(1):580. doi: 10.1038/s42003-022-03448-z, PMID: 35697829.
94. Ramdas S, Judd J, Graham S et al. (including Girotto G). “A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids”. Commun Biol. 2022 Jun 13; 5(1):580. doi: 10.1038/s42003-022-03448-z, PMID: 35697829.
95. Caiffa T, Tessitore A, Leoni L, Reffo E, Chicco D, D'Agata Mottolese B, Rubinato E, Girotto G, Lenarduzzi S, Barbi E, Bobbo M, Di Salvo G. “Long QT syndrome and left ventricular non-compaction in a family with KCNH2 mutation: A case report”. Front Pediatr. 2022 Aug 4; 10:970240. doi: 10.3389/fped.2022.970240, eCollection 2022, PMID: 35989994.
96. Young WJ, Lahrouchi N, Isaacs A, et al. (including Girotto G). “Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways”. Nat Commun. 2022 Sep 1; 13(1):5144. doi: 10.1038/s41467-022-32821-z, PMID: 36050321.
97. Mezzavilla M, Cocca M, Maisano Delser P, Badii R, Abbaszadeh F, Hadi KA, Girotto G, Gasparini P. “Ancestry-related distribution of Runs of homozygosity and functional variants in Qatari population.” BMC Genom Data. 2022 Sep 21; 23(1): 73. doi: 10.1186/s12863-022-01087-1, PMID: 36131251, PMCID: PMC9490902.
98. Tesolin P, Bertinetto FE, Sonaglia A, Cappellani S, Concas MP, Morgan A, Ferrero NM, Zabotti A, Gasparini P, Amoroso A, Quartuccio L, Girotto G. “High Throughput Genetic Characterisation of Caucasian Patients Affected by

Multi-Drug Resistant Rheumatoid or Psoriatic Arthritis." Journal of Personalized Medicine. 2022 Sep 30;12 (10):1618. doi: 10.3390/jpm12101618, PMID: 36294757, PMCID: PMC9605087.

99. Yengo L, Vedantam S, Marouli E, et al. (including Girotto G). "A saturated map of common genetic variants associated with human height". Nature. 2022 Oct; 610(7933): 704-712. doi: 10.1038/s41586-022-05275-y, Epub 2022 Oct 12, PMID: 36224396.
100. Spedicati B, Morgan A, Pianigiani G, Musante L, Rubinato E, Santin A, Nardone GG, Faletra F, Girotto G. "Challenging Occam's Razor: Dual Molecular Diagnoses Explain Entangled Clinical Pictures." Genes (Basel). 2022 Nov 3; 13 (11): 2023. doi: 10.3390/genes13112023, PMID: 36360260.
101. Aneli S, Mezzavilla M, Bortolini E, Pirastu N, Girotto G, Spedicati B, Berchialla P, Gasparini P, Pagani L. "Impact of cultural and genetic structure on food choices along the Silk Road." Proc Natl Acad Sci U S A. 2022 Nov 22; 119 (47): e2209311119. doi: 10.1073/pnas.2209311119, Epub 2022 Nov 14, PMID: 36375050.
102. Paldino A, Dal Ferro M, Stolfo D, Gandin I, Medo K, Graw S, Gigli M, Gagno G, Zaffalon D, Castrichini M, Masè M, Cannatà A, Brun F, Storm G, Severini GM, Lenarduzzi S, Girotto G, Gasparini P, Bortolotti F, Giacca M, Zacchigna S, Merlo M, Taylor MRG, Mestroni L, Sinagra G. "Prognostic Prediction of Genotype vs Phenotype in Genetic Cardiomyopathies." J Am Coll Cardiol. 2022 Nov 22; 80 (21): 1981-1994. doi: 10.1016/j.jacc.2022.08.804, PMID: 36396199.
103. D'Errico S, Bergamini PR, Fattorini P, Zanconati F, Bussani R, Cova MA, Pagnan L, Belgrano M, Gasparini P, Girotto G, Lenarduzzi S, Addobbati R, Rakar S, Aleksova A, Dal Ferro M, Zecchin M, Sinagra G. "[The Regional Registry of Sudden Cardiac Death of Friuli Venezia Giulia. Protocols, best practices and results of a multidisciplinary project]". G Ital Cardiol (Rome). 2022 Nov 23; (11):827-835. doi: 10.1714/3900.38822, PMID:36300386.
104. Kanoni S, Graham SE, Wang Y, et al. (including Girotto G). "Implicating genes, pleiotropy, and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis". Genome Biol. 2022 Dec 27; 23(1):268. doi: 10.1186/s13059-022-02837-1, PMID: 36575460.
105. Piatti G, Ambrosetti U, Aldè M, Girotto G, Concas MP, Torretta S. "Chronic Rhinosinusitis: T2r38 Genotyping and Nasal Cytology in Primary Ciliary Dyskinesia". Laryngoscope. 2023 Feb; 133(2):248-254. doi: 10.1002/lary.30112. Epub 2022 Mar 21. PMID: 35312075.
106. Di Stazio M, Zanus C, Faletra F, et al. including Girotto G. "Haploinsufficiency as a Foreground Pathomechanism of Poirer-Bienvenu Syndrome and Novel Insights Underlying the Phenotypic Continuum of CSNK2B-Associated Disorders". GENES. 2023 Feb 1. doi: 10.3390/genes14020250.
107. Lenarduzzi S, Spedicati B, Alessandrini B, Tesolin P, Paladino A, Gigli M, Sinagra G, Gasparini P, Ferro MD**,** Girotto G. "Whole exome sequencing: Clinical characterization of pedriatic and adult Italian patients affected by different forms of hereditary cardiovascular diseases'. Mol Genet Genomic Med. 2023 Feb 14; e2143. doi: 10.1002/mgg3.2143. Online ahead of print. PMID: 36788754.
108. Spedicati B, Santin A, Nardone GG, Rubinato E, Lenarduzzi S, Graziano C, Garavelli L, Miccoli S, Bigoni S, Morgan A, Girotto G. "The Enigmatic Genetic Landscape of Hereditary Hearing Loss: A Multistep Diagnostic Strategy in the Italian Population". Biomedicines. 2023 Feb 24;11(3):703. doi: 10.3390/biomedicines11030703. PMID: 36979683.
109. Bakker MK, Kanning JP, Abraham G, et al. (including Girotto G). "Genetic Risk Score for Intracranial Aneurysms: Prediction of Subarachnoid Hemorrhage and Role in Clinical Heterogeneity". Stroke. 2023 Mar; 54(3):810-818. doi: 10.1161/STROKEAHA.122.040715. Epub 2023 Jan 19. PMID: 36655558.
110. Mathieson I, Day FR, Barban N, et al. (including Girotto G). "Genome-wide analysis identifies genetic effects on reproductive success and ongoing natural selection at the FADS locus". Nat Hum Behav. 2023 Mar 2. doi: 10.1038/s41562-023-01528-6. Online ahead of print. PMID: 36864135.
111. Young WJ, Haessler J, Benjamins JW, et al. (including Girotto G). "Genetic

architecture of spatial electrical biomarkers for cardiac arrhythmia and relationship with cardiovascular disease". Nat Commun. 2023 Mar 14;14(1):1411. doi: 10.1038/s41467-023-36997-w. PMID: 36918541.

112. Reurink J, Weisschuh N, Garanto A, et al. (including Girotto G). "Whole genome sequencing for USH2A-associated disease reveals several pathogenic deep-intronic variants that are amenable to splice correction". HGG Adv. 2023 Jan 18;4(2):100181. doi: 10.1016/j.xhgg.2023.100181. eCollection 2023 Apr 13.PMID: 36785559.
113. Luppieri V, Pecori A, Spedicati B, Schito R, Pozzan L, Santin A, Girotto G, Cadenaro M, Concas MP. "Odontostomatological traits in North-Eastern Italy's isolated populations: an epidemiological cross-sectional study". J. Clin. Med. 2023 Apr 6; *12*(7), 2746. doi.org/10.3390/jcm12O72746.
114. Jonard L, Brotto D, Moreno-Pelayo MA, et al. (including Girotto G). "Genetic Evaluation of Prelingual Hearing Impairment: Recommendations of a European Network for Genetic Hearing Impairment". Audiol Res. 2023 May 10; 13(3):341-346. doi: 10.3390/audiolres13030029. PMID: 37218840.
115. Nardone GG, Spedicati B, Concas MP, Santin A, Morgan A, Mazzetto L, Battagla-Parodi M, Girotto G. "Identifying missing pieces in color vision defects: a genome-wide association study in Silk Road populations." Front Genet. 2023 Jun 9;14:1161696. doi: 10.3389/fgene.2023.1161696. eCollection 2023. PMID: 37359372.
116. 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. "Puzzling Out the Genetic Architecture of Endometriosis: Whole-Exome Sequencing and Novel Candidate Gene Identification in a Deeply Clinically Characterised Cohort". Biomedicines https://doi.org/10.3390/biomedicines11082122.
117. van de Vegte YJ, Eppinga RN, van der Ende MY, et al. (including Girotto G). "Genetic insights into resting heart rate and its role in cardiovascular disease". Nat Commun. 2023 Aug 2; 14(1): 4646. doi: 10.1038/s41467-023-39521-2. PMID: 37532724.

Abstracts

- Khalifa Al Kowari M, Gasparini P, Abdulhadi K, Siam R, Najjar N, Al-Sulaiteen M, Dipresa S, Badii R, Girotto G. "Mutations in GJB2, GJB6 and mDNA 1555 A>G variant explain only a minority of cases of nonsyndromic hearing loss in the Qatari populations". Qatar Foundation Annual Research Forum 2010, 2010 Dec 12; Doha, Qatar (Podium presentation BM04).
- Girotto G, Pirastu N, Dipresa S and Gasparini P. "Genetics of hearing loss". XV Convention Telethon, 2011 Mar 7-9; Riva del Garda, Italia (Poster presentation 208).
- Girotto G, Pirastu N, Sorice R, Biino G, Campbell H, d'Adamo AP, Hastie ND, Nutile T, Polasek O, Portas L, Rudan I, Ulivi S, Zemunik T, Wright AF, Ciullo M, Hayward C, Pirastu M, and Gasparini P. "Genetic study of hearing function, thresholds and age-related loss: a genome-wide association study in European isolated populations identifies new loci and pathways". Genomic Disorders 2011 - The Genomics of Rare Diseases, Wellcome Trust Conference Centre; Hinxton, Cambridge, Regno Unito (Poster presentation P49).
- Girotto G, Pirastu N, Sorice R, Biino G, Campbell H, d'Adamo AP, Hastie ND, Nutile T, Polasek O, Portas L, Rudan I, Ulivi S, Zemunik T, Wright AF, Ciullo M, Hayward C, Pirastu M, Gasparini P. "A genome-wide association study in European isolated populations identifies new loci and pathways for hearing function, thresholds and age-related loss". ESHG 2011; Amsterdam, Paesi Bassi (Poster presentation P08.42).
- Girotto G, Pirastu N, Lewis M, Steel KP, Gasparini P. "Genome wide association study of normal hearing function and age-related hearing loss: identification of new candidate genes". MBDH 2011; Hinxton, Cambridge, Regno Unito (Poster presentation P80).
- Gasparini P, Khalifa Al Kowari M, Girotto G, Abdulhadi K, Dipresa S, Fabretto A, Faletra F, Vozzi D, Licastro D, Siam R, Najjar N, Badii R. "High throughput technologies to study the molecular bases of heareditary hearing loss in the Italian and Qatari populations". MBDH 2011; Hinxton, Cambridge, Regno Unito (Poster presentation T25).

- Girotto G, Lewis M, Pirastu N, Licastro D, Vozzi D, Steel KP, Gasparini P. “A comprehensive approach to the study of hearing loss: identification of new candidate gene and pathways”. ICHG-ASHG 2011; Montreal, Canada (Poster presentation 612W).
- Girotto G, Pirastu N, Lewis M, Lorente B, Steel KP, Gasparini P. “Identification of new candidate genes for normal hearing function and age-related hearing loss by genome-wide association studies”. SIGU 2011; Milan, Italia (Poster presentation 20531NX).
- Girotto G, Alkowari M, Abdulhadi K, Dipresa S, Vozzi D, Licastro D, Athanasakis E, Siam R, Najjar N, Badii R, Gasparini P. “Identification of new deafness genes using high throughput technologies in Italian and Qatari population”. SIGU 2011; Milan, Italia (Podium presentation 20532XZ).
- Girotto G, Alkowari M, Abdulhadi K, Dipresa S, Vozzi D, Licastro D, Athanasakis E, Siam R, Najjar N, Badii R, Gasparini P. “Hereditary hearing loss in Italian and Qatari population: identification of new deafness genes using high throughput technologies”. Alp Nano bio International School 3, 2012 Jan 23-27; Vipiteno, Italia.
- Girotto G, Alkowari M, Abdulhadi K, Faletra F, Vozzi D, Licastro D, Athanasakis E, Badii, Gasparini P. “Identification of new deafness genes using high throughput technologies in Italian and Qatari populations”. ARO meeting 2012; San Diego, USA (Podium presentation ID 464).
- Girotto G, Pirastu N, Lewis M, Lorente B, Steel KP, Gasparini P. “Genome wide association study aimed at the identification of new candidate genes for age-related hearing loss”. ARO meeting 2012; San Diego, USA (Poster presentation ID 457).
- Girotto G, Lewis M, Buniello A, Lorente-Cánovas B, Pirastu N, Steel KP, Gasparini P. “New genes for normal hearing function and age-related hearing loss by genome-wide association and expression studies”. ESHG 2012; Nurnberg, Germania (Poster presentation P08.31).
- Girotto G, Alkowari M, Abdulhadi K, Faletra F, Vozzi D, Licastro D, Siam R, Najjar N, Badii R, Gasparini P. “High throughput technologies aimed at the identification of new candidate genes in Italian and Qatari population”. ESHG 2012; Nurnberg, Germania (Poster presentation P12.105).
- Girotto G, Vuckovic D, Buniello A, Lewis M, Lorente B, Steel KP, Gasparini P. “Genetic and environmental factors involved in normal hearing function and age-related hearing loss”. The Brain Prize Meeting 2012; Middelfart, Danimarca (Podium presentation).
- Girotto G, Vuckovic D, Buniello A, Steel KP, Gasparini P. “Genetic and environmental factors for normal hearing function and age-related hearing loss: a broad study from Europe to Central Asia”. SIGU 2012; Sorrento, Italia (Poster presentation P.035).
- Girotto G, Faletra F, Licastro D, Vozzi D, Abdulhadi K, Dipresa S, Athanasakis E, Khalifa Alkowari M, Badii R, Gasparini P. “Linkage studies and Whole Exome Sequencing: a useful combined strategy to identify new deafness genes”. SIGU 2012; Sorrento, Italia (Podium presentation 22538FD).
- Girotto G, Vuckovic D, Biino G, Panu F, Pirastu M, Gasparini P. “Environmental and lifestyle factors involved in normal hearing function and age-related hearing loss”. ARO meeting 2013; Baltimore, USA (Poster presentation 985).
- Girotto G, Vuckovic D, Buniello A, Lewis M, Lorente B, Steel KP, Gasparini P. “Genome wide association analysis and expression studies involved in hearing function and age-related hearing loss”. ARO meeting 2013; Baltimore, USA (Podium presentation 992).
- Girotto G, Faletra F, Licastro D, Vozzi D, Abdulhadi K, Dipresa S, Athanasakis E, Khalifa Alkowari M, Badii R, Gasparini P. “Linkage studies and Whole Exome Sequencing analysis aimed at the identification of new deafness genes”. ARO meeting 2013; Baltimore, USA (Poster presentation 1004).
- Girotto G, Vuckovic D, Buniello A, Lewis M, Steel KP, Paolo Gasparini. “Genetic and environmental factors involved in normal hearing function and age-related hearing loss”. ESHG 2013; Parigi, Francia (Poster presentation P16.049).
- Vozzi D, Girotto G, Faletra F, Abdulhadi K, Vuckovic D, D’Eustacchio A, Khalifa Alkowari M, Badii R, Gasparini G. “Identification of new genes for

Hereditary Hearing Loss (HHL) using linkage studies and Whole Exome Sequencing analysis". ESHG 2013; Parigi, France (Podium presentation C14.3).

- Cocca M, Traglia M, Memari Y, Walter K, Sala C, Bomba L, Girotto G, Masciullo C, Barbieri C, D'Adamo P, Gasparini P, Soranzo N, Toniolo D. "A whole-genome reference panel enriched in Italian lower-frequency Variants". ESHG 2013; Parigi, Francia (Poster presentation P12.02).
- Girotto G, Vuckovic D, Buniello A, Lewis M, Steel KP, Gasparini P. "A complex strategy for complex traits: identification of new genes involved in hearing function and age-related hearing loss". MBHD 2013; Stanford USA (Podium presentation).
- Nolan L, Maier H, Borgmeyer-Hermans I, Girotto G, Ecob R, Pirastu N, Cadge B.A, Hubner C, Gasparini P, Strachan DP, Davis A, Dawson SJ. "Genetic variation in ESRRG is associated with adult hearing status in humans and its targeted deletion causes hearing loss in mice". MBHD 2013; Stanford USA (Podium presentation).
- Girotto G, Faletra F, Vozzi D, Vuckovic D, Abdulhadi K, Khalifa Alkowari M, Badii R, Gasparini P. "Linkage studies and Whole Exome Sequencing analysis aimed at the identification of new Hereditary Hearing Loss genes/loci". MBHD 2013; Stanford USA (Poster presentation).
- Vozzi D, Morgan A, Morgutti M, Lenarduzzi S, Rubinato E, Faletra F, Girotto G, Gasparini P. "Targeted Exome Capture and Paired-End Massively Parallel Sequencing Reveals New Mutations for Human Hereditary Deafness in the Middle East". MBHD 2013; Stanford USA (Poster presentation).
- Di Stazio M, Vozzi D, Girotto G, Faletra F, Vuckovic D, Gasparini P. "A Y-linked form of Hereditary Hearing Loss in an Italian Family". MBHD 2013; Stanford USA (Podium presentation).
- Vozzi D, Girotto G, Faletra F, Rubinato E, Morgan, A Vuckovic D, Lenarduzzi S, Morgutti M, Gasparini P. "Next Generation Sequencing (NGS) approaches to identify Hereditary Hearing Loss (HHL) Genes, to Improve Diagnosis and Prevention of Deaf Patients". SIGU 2013; Roma, Italia (Podium presentation).
- Vozzi D, Girotto G, Faletra F, Rubinato E, Morgan A, Vuckovic D, Lenarduzzi S, Morgutti M, Gasparini P. "Shouting won't help: a combined strategy (Next Generation Sequencing and Linkage analysis) to identify Hereditary Hearing Loss (HHL) genes in affected families". ASHG 2013; Boston, USA (Poster presentation).
- Girotto G, Vuckovic D, Buniello A, Steel KP, Gasparini P. "Hearing function and loss: "A complex multistep strategy" to identify genetics and environmental factors". ASHG 2013; Boston, USA (Poster presentation 1723F).
- Girotto G, Vuckovic D, Concas M, Nutile T, Ciullo M, Pirastu M, Gasparini P. "Genome-wide association study, "Genome-Wide Association Study, Whole Genome Sequencing and Animal Models to Study Normal Hearing Function and AgeRelated Hearing Loss Giorgia Girotto". ARO Meeting 2014; San Diego, USA (Poster presentation PS - 284).
- Gasparini P, Vozzi D, Morgan A, Vuckovic D, Rubinato E, Di Stazio M, Faletra F, Lenarduzzi S, Girotto G. "The Use of Different NGS Protocols to Study Inherited Forms of Hereditary Hearing Loss (HHL)." ARO Meeting 2014; San Diego, USA (Podium presentation PD – 008).
- Vuckovic D, Girotto G, Gasparini P. "Screening of 250.000 DNA Functional Variants in Large Cohorts of Age-Related Hearing Loss Patients and Normal Hearing Subjects." ARO Meeting 2014; San Diego, USA (Poster presentation PS – 285).
- Bowl M, Aguilar C, Morse S, Dorning J, Shanthakumar P, King R, Chessum L, Wisby L, Simon M, Parker A, Girotto G, Dawson S, Gasparini P, Wells S, Potter P, Brown S. "The Harwell Aging Mutant Screen Identifies Novel Models of Age-Related Hearing Loss". ARO Meeting 2014; San Diego, USA (Podium presentation PD – 131).
- Vuckovic D, Dawson S, Concas MP, Ciullo M, Nutile T, Cappellani S, Pirastu M, Gasparini P, Girotto G. "Hearing and ageing: a complex genomic strategy leading to new genes/variants identification in European and Central Asia populations". ESHG 2014; Milano, Italia (Poster presentation P02.10-M).
- Girotto G, Vozzi D, Rubinato E, Morgan E, Abdulhadi K, Vuckovic D, Di Stazio

M, D'Eustacchio A, La Bianca M, Badii R, Gasparini P. "New Hereditary hearing loss (HHL) genes/mutations identified by High throughput sequencing and genotyping in the Italian and Qatari populations". ESHG 2014; Milano, Italia (Podium presentation C12.2).

- Lenarduzzi S, Graziano C, Castorina P, Ambrosetti U, Morgan A, Rubinato E, Vozzi D, Girotto G, Morgutti M. "Targeted re-sequencing and USH syndrome: a robust and accurate protocol overcoming the problem of genetic heterogeneity and leading to the discovery of several new mutations". ESHG 2014; Milano, Italia (Poster presentation P14.90-M).
- Girotto G, Dawson S, Concas MP, Ciullo M, Nutile T, Cappellani S, Loukola A, Bahl A, Pirastu M, Gasparini P, Vuckovic D. "Hearing and ageing: a multistep genomic strategy to identify new genes/variants in European and Central Asian populations". IEB 2014; Sheffield, Regno Unito (Podium presentation LS4-015).
- Girotto G, Vozzi D, Rubinato E, Morgan A, Abdulhadi K, Vuckovic D, Di Stazio M, D'Eustacchio A, La Bianca M, Badii R, Gasparini P. "New Hereditary hearing loss (HHL) genes/mutations identified by High throughput technologies in the Italian and Qatari populations". IEB 2014; Sheffield, Regno Unito (Podium presentation LS7-028).
- Girotto G, Vozzi D, Badii R, Rubinato E, Morgan A, Vuckovic D, Khalifa Alkowari M, D'Eustacchio A, La Bianca M, Gasparini P, Abdulhadi K. "New Hereditary Hearing Loss (HHL) genes/mutations identified by High throughput technologies in the Qatari population". ARC'14, 2014 Nov 18-19; Doha, Qatar (Podium presentation).
- Girotto G, Morgan A, Vuckovic D, Rubinato E, Vozzi D, Gandin I, Di Stazio M, Badii R, Vozzi D, Gasparini P. "Hereditary Hearing Loss: the use of a high throughput and multistep approach to identify new genes". ARO meeting 2015; Baltimore, USA (Podium Presentation PD-83 by G.Girotto).
- Gasparini P, Vuckovic D, Girotto G. "Genetics of Age-related Hearing Loss". ARO meeting 2015; Baltimore, USA (Poster presentation).
- Morgan A, Vozzi D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G (PD-84). "A New Targeted Re-Sequencing Protocol to Investigate the Genetic Causes of Age-Related Hearing Loss". ARO meeting 2015; Baltimore, USA (Podium Presentation PD-84 by A.Morgan).
- Girotto G, Scheffer DI, Morgan A, Vozzi D, Rubinato E, Di Stazio M, Muzzi E, Pensiero S, Giersch AB, Corey DP, Gasparini P. "NGS revealed PSIP1/LEDGF as a new gene causing sensorineural progressive hearing loss and variable eye phenotypes". ESHG 2015; Glasgow, Regno Unito (Poster presentation PM02.30).
- Vuckovic D, Concas MP, La Bianca M, Pirastu M, Gasparini P, Girotto G. "Multi-Trait Genome-wide Association Studies (GWAS) to investigate hearing function and interactions with BMI and blood pressure." ESHG 2015; Glasgow, Regno Unito (Poster presentation PM02.22).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G. "Genome-wide association study and targeted re-sequencing: a new combined approach to investigate the genetic causes of Age-related Hearing Loss (ARHL)". ESHG 2015; Glasgow, Regno Unito (Poster presentation PM02.50).
- Girotto G, Scheffer DI, Morgan A, Vozzi D, Rubinato E, Di Stazio M, Muzzi E, Pensiero S, Giersch AB, Corey DP, Gasparini P. "Next generation sequencing for the study of hereditary hearing loss: PSIP1/LEDGF as a new gene causaing sensorineural progressive hearing loss and variable eye phenotypes". IEB 2015; Roma, Italia (Podium Presentation O22 by G.Girotto).
- Orzan E, Girotto G, Gregori M, Marchese C, Monasta L, Morgan A, La Bianca M, Vozzi D, Gasparini P. "Diagnostic yield of a targeted gene sequencing approach within a regional universal newborn hearing screening and childhood surveillance program: a 2-year experience in Friuli Venezia Giulia". IEB 2015; Roma, Italia (Poster presentation P85).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G. "A new targeted re-sequencing panel for unveiling the genetic causes of age-related hearing loss (ARHL)". IEB 2015; Roma, Italia (Poster presentation P86).
- Girotto G, Scheffer DI, Morgan A, Vozzi D, Rubinato E, Di Stazio M, Muzzi E, Pensiero S, Giersch AB, Corey DP, Gasparini P. "Whole Exome sequencing for

the study of hereditary hearing loss: PSIP1/LEDGF as a new gene causing sensorineural progressive hearing loss and a variable eye phenotype." Genetika 2015; Rogaška Slatina, Slovenia (Podium presentation Section 1-Genetic diseases I, oral communication by Girotto G).

- Girotto G, Morgan A, Vuckovic D, Rubinato E, Di Stazio M, Badii R, Al-Kowary M, Vozzi D, Gasparini P. "Next generation sequencing and animal studies: a multistep approach for the discovery of new Hereditary Hearing Loss genes". ARO meeting 2016; San Diego, USA (Podium Presentation PD25 by Girotto G).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G. "Genome Wide Association Studies (GWAS), Targeted Re-Sequencing (TRS) and Animal Studies: a combined approach aimed to investigate the genetic causes of Age-related Hearing Loss (ARHL)". ARO meeting 2016; San Diego, USA (Podium Presentation PD29 by Morgan A).
- Rubinato E, Azaiez H, BlackZiegelbein E, Morgan A, Booth K, Vozzi D, Sloan C, Frees K, Vuckovic D, Concas MP, Ephraim S, Biino G, Vaccargiu S, Girotto G, Pirastu M, Gasparini P, Smith R. "High Frequency of GJB2 Splice-site Mutation c.-22-2A>C in a Large Cohort of Italian Age Related Hearing Loss Patients and Matched Controls". ARO meeting 2016; San Diego, USA (Poster Presentation PS568).
- Khalifa Alkowari M, Morgan A, Vozzi D, Vuckovic D, Rubinato E, La Bianca M, Baghat S, Badii R, Abdulhadi K, Gasparini P, Girotto G. "Hereditary Hearing Loss in Qatar: a powerful strategy to detect new genes". ARC'16, 2016; Doha, Qatar (Poster Presentation).
- Morgan A, Khalifa Alkowari M, Baghat S, Rubinato E, Vuckovic D, Badii R, Gasparini P, Abdulhadi K, Vozzi D, Girotto G. "Hereditary Hearing Loss (HHL) genes discovery in the Qatari population". PAHGC|6th Pan Arab Human Genetics Conference 2016; Dubai, Emirati Arabi Uniti (Poster Presentation).
- Bhagat S, Vozzi D, Morgan A, Girotto G, Logendra B, Alkowari Moza K, Abdulhadi K., Badii R, Gasparini P, Al-Nabet A. "Targeted Next-Generation Sequencing for Molecular Diagnosis of Non-syndromic Hearing Loss in Qatar". ARC'16, 2016; Doha, Qatar (Poster Presentation).
- Vuckovic D, Morgan A, Concas MP, Ciullo M, Nutile T, Pirastu M, Huentelman MJ, Schrauwen I, Friedman RA, Fransen E, Van Camp G, Gasparini P, Girotto G. "Unravelling human complex traits: the case of hearing function and age related hearing loss". ESHG 2016; Barcelona, Spagna (Poster presentation P02.09).
- Espino-Guarch M, Prat E, Font-Llitjós M, Errasti E, Girotto G, Vilches C, Celaya AM, Gónzalez L, Vuckovic D, Sahún I, Murillo-Cuesta S, López de Heredia M, Bodoy S, Dierssen M, Varela-Nieto I, Gasparini P, Palacín M, Nunes V. "Mutations in the amino acid transporter LAT2 (SLC7A8) cause agerelated hearing loss". ESHG 2016; Barcelona, Spagna (Poster presentation P02.02).
- Morgan A, Vozzi D, Rubinato E, Khalifa AlKowari M, Di Stazio M, Badii R, Abdulhadi K, La Bianca M, Gasparini P, Girotto G. "Targeted/Whole Exome Sequencing plus Animal Models for the molecular characterisation of Hereditary Hearing Loss (HHL)". ESHG 2016; Barcelona, Spain (Poster presentation P02.23).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G. "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)". MBHD 2016; Hinxton, Cambridge, Regno Unito (Poster presentation P58).
- Rubinato E, Morgan A, Vozzi D, Vuckovic D, Distazio M, Ambrosetti U, Castorina P, Gasparini P, Girotto G. "Next generation sequencing and functional studies for the discovery of new Hereditary Hearing Loss (HHL) genes in Italian families and isolated patients". MBHD 2016; Hinxton, Cambridge, Regno Unito (Poster presentation).
- Alkowari MK, Bhagat S, Morgan A, Hayder Y, Logendra B, Najjar N, Gasparini P, Badii B, Abdulhadi K, Vozzi D, Girotto G. "Molecular and functional high throughput characterization of hearing loss in the Qatari population". MBHD 2016; Hinxton, Cambridge, Regno Unito (Poster presentation).
- Girotto G, Morgan A, Vozzi D, Rubinato E, AlKowari MK, Di Stazio M, Badii R, Bhaghat S, Abdulhadi K, La Bianca M, Gasparini P. "Next generation sequencing (NGS) and in vivo/in vitro approaches for the molecular characterisation of

Hereditary Hearing Loss in Italian and Qatari families". Inner Ear Biology 2016; Montpellier, Francia (Podium Presentation 032 by Girotto G.).

- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Concas MP, Pirastu M, Gasparini P, Girotto G. "Genome wide association studies (GWAS), targeted re-sequencing (TRS) and animal studies: A powerful multi-step approach for the discovery of the genetics of age-related hearing loss (ARHL)." Inner Ear Biology 2016; Montpellier, Francia (Podium Presentation 033 by Morgan A.).
- Girotto G, Morgan A, Vozzi D, Rubinato E, K.AlKowari M, Di Stazio M, Badii R, Bhaghat S, Abdulhadi K, La Bianca M, Castorina P, Ambrosetti U, Gasparini P. "Next generation sequencing (NGS) and in vitro/in vivo approaches for the molecular characterization of Italian and Qatari families affected by hereditary hearing loss". SIGU meeting 2016; Torino, Italia (Podium Presentation by Girotto G.).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Brumat M, Ambrosetti U, Gasparini P, Girotto G. "Combining genome wide association studies (GWAS), targeted re-sequencing (TRS) and animal studies for the discovery of the genetics of age-related hearing loss (ARHL)". SIGU meeting 2016; Torino, Italia (Podium Presentation by Morgan A.).
- Morgan A, Vozzi D, Vuckovic D, La Bianca M, D'Eustacchio A, Brumat M, Ambrosetti U, Gasparini P, Girotto G. "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". ARO Meeting 2017; Baltimore, USA (Poster presentation PS124).
- Girotto G, Morgan A, Vozzi D, Rubinato E, Alkowari M, Di Stazio M, Badii R, Bhagat S, Abdulhadi K, Ambrosetti U, La Bianca M, Gasparini P. "Next Generation Sequencing (NGS) and Functional Studies for the Molecular Characterisation of Families and Sporadic Cases Affected by Hereditary Hearing Loss". ARO Meeting 2017; Baltimore, USA (Poster presentation PS121).
- Vuckovic D, Morgan A, Brumat M, Cocca M, Catamo E, Ambrosetti U, Gasparini P, Girotto G. "Whole Genome Sequencing (WGS) Reveals Cumulative Variation Effects in Age Related Hearing Loss (ARHL)". ARO Meeting 2017; Baltimore, USA (Poster presentation PS120).
- Morgan A, Vuckovic D, Di Stazio M, Alkowari M, Badii R, Abdulhadi K, La Bianca M, Ambrosetti U, Gasparini P, Girotto G. "Next Generation Sequencing (NGS) followed by in vitro and in vivo functional studies revealed new genes for both Hereditary (HHL) and Age Related Hearing Loss (ARHL)". ESHG 2017; Copenhagen, Danimarca (Poster presentation P02.01A).
- Vuckovic D, Mezzavilla M, Cocca M, Morgan A, Brumat M, Gasparini P, Girotto G. "Genome wide sequencing (WGS) reveals new insights into Age Related Hearing Loss (ARHL): cumulative effects and the role of selection". ESHG 2017; Copenhagen, Danimarca (Poster presentation P02.07C).
- Cocca M, Mezzavilla M, Barbieri C, Brumat M, Concas M, Vuckovic D, Robino A, Gandin I, Girotto G, Sala C, Gasparini P, Soranzo N, Toniolo D. "Genomic characterization of three Italian isolated populations through low-coverage whole genome sequencing". ESHG 2017; Copenhagen, Danimarca (Poster presentation P1834B).
- Girotto G, Morgan A, Lenarduzzi S, Morgutti M, La Bianca M, Cappellani S, Faletra F, Sirchia F, Graziano C, Orzan E, Pecile V, Gasparini P. "The Italian map of hereditary hearing loss genes: a peculiar case of Uniparental Disomy (UPD) and the discovery of new genes". SIGU 2017; Napoli, Italia (Podium presentation by G. Girotto).
- Morgan A, Distazio M, La Bianca M, D'Eustacchio A, Brumat M, Ambrosetti U, Gasparini P, Girotto G. "SLC9A3R1: a new gene involved in human age-related hearing loss (ARHL)". SIGU 2017; Napoli, Italia (Podium presentation by A. Morgan)
- Black-Ziegelbein E, Azaiez H, Rubinato E, Wang K, Morgan A, Booth K, Vozzi D, Vuckovic D, Concas MP, Biino G, Vaccargiu S, Pirastu M, Girotto G, Gasparini P, Smith R. "Haplotype Reconstruction Provides Insights into the Contributions of known Deafness-Causing Genes to Age-Related Hearing Loss". ARO meeting 2017; Baltimore, USA (Poster presentation).
- Ingham NJ, Rook V, Buniello A, Girotto G, Gasparini P, Steel KP. "Analysis of

candidate genes for age-related hearing loss from Genome-wide Association Studies". ARO meeting 2017; Baltimore, USA (Poster presentation).

- Girotto G, Morgan A, Brumat M, Di Stazio M, Cappellani S, Campana E, Ambrosetti U, La Bianca M, Orzan E, Gasparini P. "Discovery of new Hereditary Hearing Loss (HHL) genes by Whole Exome Sequencing (WES) and in vitro/in vivo functional studies: five years of experience". ESHG 2018; Milano, Italia (Poster presentation P02.58A/A).
- Lenarduzzi S, Morgan A, Cappellani S, Pecile V, Morgutti M, Orzan E, Ambrosetti U, La Bianca M, Faletra F, Grosso E, Sirchia F, Sensi A, Graziano C, Seri M, Gasparini P, Girotto G. "Targeted Re-Sequencing (TRS) and high density SNP array for the molecular characterisation of Hereditary Hearing Loss (HHL)". ESHG 2018; Milano, Italia (Poster presentation P02.22A/A).
- Morgan A, Brumat M, Cocca M, Di Stazio M, Bassani S, La Bianca M, Gasparini P, Girotto G. "NGS and animal model reveal SLC9A3R1 as a new gene involved in human age-related hearing loss (ARHL)". ESHG 2018; Milano, Italia (Podium presentation C23.2 by A. Morgan).
- Steel KP, Ingham NJ, Lewis M, Buniello A, Pearson S, White J, Girotto G, Gasparini P, Schulte B, Dubno J, Dawson S. "Molecular pathways underlying progressive hearing loss". MBHD 2018; Gottingen, Germania (Podium presentation by K. Steel).
- Aguilar C, Boucher S, Parker A, Codner G, Girotto G, Wells S, Gasparini P, Dawson S, Teboul L, Brown S, Bowl M. "Investigating the genetic basis of age-related hearing loss - Human GWAS to mouse models". MBHD 2018; Gottingen, Germania (Poster presentation N°56).
- Girotto G, Brumat M, Morgan A, Campana E, Cocca M, Catamo E, La Bianca M, Ambrosetti U, Gasparini P. "Genome-wide association studies (GWAS), targeted re-sequencing (TRS) and in vitro functional validations for the study of normal hearing function (NHF) and age-related hearing loss (ARHL)". MBHD 2018; Gottingen, Germania (Podium presentation by G. Girotto).
- Abdulhadi K, Girotto G, Mezzavilla M, Alkowari M, Di Stazio M, Morgan A, Cocca M, Hayder Y, Logendra B, Najjar N, Paolo Gasparini, Badii R. "Genetic insights of age-related hearing loss in Qatar." MBHD 2018; Gottingen, Germania (Poster presentation N°24).
- Bassani S, Girotto G, Abdulhadi K, Alkowari M, Morgan A, La Bianca M, Badii R, Gasparini P. "Hereditary Hearing Loss (HHL) in Qatar: targeted re-sequencing (TRS) and whole exome sequencing (WES) for the discovery of new mutations and genes". MBHD 2018; Gottingen, Germania (Poster presentation N°26).
- Morgan A, Lenarduzzi S, Cappellani S, Pecile V, Morgutti M, Orzan E, Ghiselli S, Ambrosetti U, La Bianca M, Faletra F, Grosso E, Sirchia F, Sensi A, Graziano C, Seri M, Gasparini P, Girotto G. "Genomic analysis of a large cohort of Hereditary Hearing Loss (HHL) Italian patients identified new genes/alleles and revealed the important role of copy number variants (CNVs) in hearing phenotype, in addition to the discovery of the first case of uniparental disomy (UPD) in LOXHD1 gene." MBHD 2018; Gottingen, Germania (Poster presentation n°44).
- Mezzavilla M, Cocca M, Barbieri C, Concas MP, Gandin I, Brumat M, Robino A, Trudu M, Sala C, Vuckovic D, Matullo G, Ozren P, Girotto G, Soranzo N, Toniolo D, Gasparini P. "A first view on the Italian Genome Reference". SIGU meeting 2018; Catania, Italia.
- Girotto G, Brumat M, Morgan A, Campana E, Cocca M, La Bianca M, Di Stazio M, Ambrosetti U, Gasparini P. "Genome-wide association studies (GWAS), targeted re-sequencing (TRS) and functional studies for the study of age-related hearing loss (ARHL)" SIGU meeting 2018; Catania, Italia (Podium presentation by G.Girotto).
- Morgan A, Lenarduzzi S, Cappellani S, Pecile V, Morgutti M, Ambrosetti U, La Bianca M, Faletra F, Grosso E, Sirchia F, Sensi A, Graziano C, Seri M, Gasparini P, Girotto G. "Targeted Re-Sequencing (TRS), High-Density (HD) SNP arrays and Whole Exome Sequencing (WES) for the molecular characterisation of Hereditary Hearing Loss (HHL)". SIGU meeting 2018; Catania, Italia (Podium presentation by A. Morgan).
- Morgan A., Lenarduzzi S., Cappellani S., Faletra F., Koboldt D., Dell'Orco D., Paolo Gasparini P., Girotto G. "Genomic analysis of a large cohort of Hereditary Hearing Loss (HHL) Italian patients: the usefulness of multiple integrated

methodologies for the identification of new genes/alleles" Genomics of rare disease 2019

- Cocca M., Faletra F., Girotto G., Morgan A., Sirchia F., Spedicati B., Palmisano R., Barbieri C., Toniolo D., Gasparini P. "The relevant role of Italian genetic isolated for the study of Human Knockouts (HKO)". Genomics of rare disease 2019.
- Morgan A, Koboldt D, Barrie E, Crist E, García G, Mezzavilla M, Faletra F, Mosher T, Wilson R, Blanchet C, Manickam K, Roux AF, Gasparini P, Dell'Orco D, Girotto G. "Mutations in PLS1, encoding fimbrin, cause autosomal dominant non-syndromic hearing loss (ADNSHL)". ESHG 2019.
- Brumat M., Morgan A., Cocca M., Mezzavilla M., Toniolo D., Van Camp G., Biino G., Ambrosetti U., Ghiselli S., Gasparini P., Girotto G. "Preliminary results from a new GWAS meta-analysis point at new loci for age-related hearing loss (ARHL)". ESHG 2019.
- Morgan A., Lenarduzzi S., Cappellani S., Faletra F., Koboldt, Dell'Orco D., Gasparini P., Girotto G. "Genomic analysis of a large cohort of Hereditary Hearing Loss (HHL) Italian patients: the usefulness of multiple integrated methodologies for the identification of new genes/alleles." Genomics of rare disorders 2019.
- Cocca M, Faletra F, Girotto G, Morgan A, Sirchia F, Spedicati B, Palmisano R, Barbieri C, Toniolo D, Gasparini P. "The relevant role of Italian genetic isolated for the study of Human Knockouts (HKO)." Genomics of rare disorders 2019; Hinxton, Cambridge, Regno Unito.
- Morgan A, Koboldt DC, Barrie ES, Crist ER, García G, Mezzavilla M, Faletra F, Mihalic Mosher T, Wilson RK, Blanchet C, Manickam K, Roux AF, Gasparini P, Dell'Orco D, Girotto G. "Mutations in PLS1 cause autosomal dominant non-syndromic hearing loss in three families of European ancestry". TL4 Oral communication, IEB meeting 2019; Hinxton, Cambridge, Regno Unito.
- Ghiselli S, Castro V, Gregori M, Bruno I, Marchi R, Pensiero S, Bin M, Faletra F, Girotto G, Gasparini P, Orzan E. "The complexity of multidisciplinary evaluation of children with sensorineural hearing loss: three case reports". P5 IEB meeting 2019; Padova, Italia.
- Fetoni AR, Zorzi V, Paciello F, Ziraldo G, Raspa M, Scavizzia F, Salvatore AM, Crispino G, Tognola G, Gentile G, Spampinato AG, Cuccaro D, Guarnaccia M, Morello G, Van Camp G, Fransen E, Brumat M, Girotto G, Paludetti G, Gasparini P, Cavallaro S, Mammano F. "Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway". IEB meeting 2019 P44; Padova, Italia.
- Brumat M, Morgan A, Cocca M, Mezzavilla M, Toniolo D, Van Camp G, Fransen E, Biino G, Ambrosetti U, Ghiselli S. Gasparini P, Girotto G. "GWAS meta-analysis points to new loci for normal hearing function and age-related hearing loss". IEB meeting 2019 P57; Padova, Italia.
- Orzan E, Feresin A, Hamade M, Gregori M, Castro V, Grasso DL, Cossu P, Tonon E, Gasparini P, Girotto G, Faletra F, Bruno I, Travan L, Ghiselli S. "From hearing screening into a systematic etiologic approach and follow-up of Paediatric Permanent Hearing Loss in Friuli-Venezia Giulia Region". IEB meeting 2019 P73; Padova, Italia.
- Girotto G, Morgan A, Koboldt DC, Barrie ES, Crist ER, García García G, Mezzavilla M, Faletra F, Mihalic Mosher T, Wilson RK, Blanchet C, Manickam K, Roux AF, Dell'Orco D, Gasparini P. "PLS1, encoding plastin-1, causes autosomal dominant (AD) non-syndromic hearing loss in three families of European ancestry". SIGU 2019, 2019 Nov 13-16; Roma, Italia (Podium Presentation C009).
- Spedicati B, Palmisano R, Cocca M, Barbieri C, Sirchia F, Mezzavilla M, Morgan A, Faletra F, Gasparini P, Girotto G. "We are all experiments of nature: the fascinating role of Human Knockouts (HKO)". SIGU 2019, 2019 Nov 13-16; Roma, Italia (Podium Presentation C020).
- Bassani S, Morgan A, Cocca M, Voisin N, Chrast J, Pradervand S, Chatron N, Delprat B, Rossel M, Locher H, Van Beelen E, Tangui M, Gasparini P, Girotto G, Reymond A. "Molecular Assays, Immunohistology and Animal Models associate USP48 to Hereditary Hearing Loss". MBHD 2020; Iowa, USA.
- Brumat M, Morgan A, Nagtegaal AP, Oosterloo BC, Seshadri S, Heard-Costa NL,

Van Camp G, Fransen E, Gasparini P, Girotto G. "The G-EAR Consortium study: new candidate genes for normal hearing function (NHF) and age-related hearing loss (ARHL) in a large and deeply phenotyped cohort". ESHG 2020; Berlino, Germania.

- Robino A, Girotto G, Ambrosetti U, Piatti G. "The intriguing role of bitter taste receptor TAS2R38 in primary ciliary dyskinesia PCD". ESHG 2020; Berlino, Germania.
- Concas MP, Battistuzzi T, Cocca M, Spedicati B, Pelliccione G, Gasparini P, Girotto G. "Impairments of smell perception: the role of sex, age and genes". ESHG 2020; Berlino, Germania.
- Gasparini P, Pelliccione G, Faletra F, Dell'Orco D, Brumat M, Girotto G, Morgan A. "Identification of New Mutations and Genes by a Multi-Step Approach in more than 700 Italian Patients Affected by Hearing Loss". MBHD 2020; Iowa, USA.
- Morgan A, Pelliccione G, Faletra F, Dell'Orco D, Brumat M, Gasparini P, Girotto G. " Genomic approaches for the analysis fo more than 700 Italian patients affected by Hereditary Hearing Loss (HHL) reveal new genes/alleles". ESHG 2020; Berlino, Germania.
- Morgan A, Di Stazio M, Brumat M, Bassani S, Dell'Orco D, Marino V, Garagnani P, Giuliani C, Gasparini P, Girotto G. "Next-generation sequencing followed by in silico and in vitro functional studies revealed new candidate genes for Age Related Hearing Loss (ARHL)." MBHD 2020; Iowa; USA.
- Concas MP, Brumat M, Robino A, Catamo E, Girotto G, Mezzavilla M, Gasparini P. "Aging and sensory decays: the role of genetica and environmental factors in Italian samples." Eurosense 2020; Rotterdam, Paesi Bassi.
- Morgan A, Faletra F, Lenarduzzi S, La Bianca M, Pelliccione G, Spedicati B, Feresin A, Mazzà D, Sensi A, Graziano C, Seri M, Ambrosetti U, Gasparini P, Girotto G. "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)." SIGU meeting 2020 (online).
- Concas MP, Francescatto M, Battistuzzi T, Spedicati B, Pelliccione G, Morgan A, Feresin A, Cocca M, Gasparini P, Girotto G. "The role of bi-allelic Loss of Function variants in Olfactory Receptor genes on the perception of smell." SIGU meeting 2020 (online).
- Concas MP, Brumat M, Catamo E, Girotto G, Gasparini P. "Aging and sensory decays: the role of genetic and environmental factors in Italian samples." SIGU meeting 2020 (online).
- Spedicati B, Bosio B, D'Adamo AP, Cappellani S, Feresin A, Gasparini P, Girotto G, Faletra F, Ardisia C. "When mosaicism deceives the eye: an incidental diagnosis of Beckwith-Wiedemann syndrome". SIGU meeting 2020 (online).
- Girotto G, Spedicati B, Romano C, Garavelli L, Dentici M, Specchio N, Alfieri P, Grammatico P, Trimarchi G, Tartaglia M, Gasparini P, Faletra F, Feresin A. "The role of POGZ in neurodevelopmental disorders: from molecular diagnosis to a possible therapeutic approach". SIGU meeting 2020 (online).
- Morgan A, Faletra F, Lenarduzzi S, La Bianca M, Pelliccione G, Spedicati B, Feresin A, Mazzà D, Sensi A, Graziano C, Seri M, Ambrosetti U, Gasparini P, Girotto G. "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". ESHG 2021 (online).
- Girotto G, Cocca M, Morgan A, Catamo E, Tesolin P, Feresin A, Gasparini P, Concas M. "Genetic and environmental factors influencing sensory decays during aging in a large Italian cohort". ESHG 2021 (online).
- Tesolin P, Concas M, Cocca M, Francescatto M, Luglio A, Spedicati B, Feresin A, Morgan A, Gasparini P, Girotto G. "Human knockouts of olfactory receptors genes and smell perception impairment in a large Italian cohort". ESHG 2021(online).
- Concas M, Minelli A, Aere S, Serra F, Morgan A, Spedicati B, Morgante G, Cocca M, Gennarelli M, Gasparini P, Girotto G. "Genetic dissection of Cloninger's Temperament and Character Inventory, TCI, in an Italian isolate". ESHG 2021 (online).
- Spedicati B, Morgan A, Bonati MT, Severi G, Feresin A, Graziano C, Gasparini P,

Faletra F, Girotto G. “There’s more than meets the eye: dual molecular diagnosis in complex hearing loss patients” ESHG 2021 (online).

- Morgan A, Faletra F, Lenarduzzi S, La Bianca M, Pelliccione G, Spedicati B, Feresin A, Mazzà D, Sensi A, Graziano C, Seri M, Ambrosetti U, Gasparini P, Girotto G. “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”. ARO meeting 2021 (online).
- Concas MP, Minelli A, Aere S, Morgan A, Tesolin P, Cocca M, Gennarelli M, Gasparini P, Girotto G. “Genetics of Cloninger's Temperament and Character Inventory (TCI): results from an Italian isolate”. SIGU meeting 2021(online).
- Morgante G, Bonati MT, Faletra F, Feresin A, Luglio A, Morgan A, Musante L, Rubinato E, Spedicati B, Girotto G, Gasparini P. “A novel PRKAR1B variant as a cause of intellectual disability and hyperphagia”. SIGU meeting 2021 (online).
- Luglio A, Bonati MT, Di Bella D, Magri S, Sarto E, Nanetti L, Faletra F, Feresin A, Morgan A, Morgante G, Musante L, Rubinato E, Spedicati B, Girotto G, Taroni F, Gasparini G. “When short stature is not a small thing: a medical history that began with GH deficiency hypostaturism”. SIGU meeting 2021 (online).
- Morgan A, Faletra F, La Bianca M, Severi G, Graziano C, Gasparini P, Girotto G. “Clinical application of Whole Exome Sequencing (WES): identification of dual molecular diagnoses in patients affected by Hearing Loss (HL)”. SIGU meeting 2021(online).
- Spedicati B, Feresin A, Musante L, Morgan A, La Bianca M, Faletra F, Bonati MT, Rubinato E, Girotto G, Gasparini P. “Whole Exome Sequencing in the pediatric emergency setting: when time matters for patients’ treatment, care and management”. SIGU meeting 2021 (online).
- Serra F, Mazzetto L, Cocca M, Gasparini P, Concas MP, Girotto G. “GWAS analysis led to the identification of genes potentially involved in Color Vision Defects (CVDs) pathogenesis”. SIGU meeting 2021 (online).
- Feresin A, Spedicati B, Morgante G, Luglio A, Suergiu S, La Bianca M, Pelliccione G, Fantasia I, Stampalija T, Murru FM, Morgan A, Rubinato E, Bonati MT, Faletra F, Girotto G, Bussani R, Gasparini PG. “Whole Exome Sequencing for fetuses with structural anomalies: which contribute in the post-mortem diagnostic pathway?” SIGU meeting 2021(online).
- Feresin A, Cappellani S, Ulivi S, Spedicati B, Morgante G, Luglio A, Rubinato E, Bonati MT, Faletra F, Girotto G, Stampalija T, d’Adamo AP, Gasparini P. “An X-linked dominant condition with incomplete penetrance? A familiar case of SOX3 duplication within three generations of healthy subjects and affected fetuses”. SIGU meeting 2021 (online).
- Lenarduzzi S, Dal Ferro M, Paldino A, Alessandrini B, Sinagra G, Gasparini P, Girotto G. “Cardiomyopathies (CMs): from clinical characterization to genetic analysis in patients recruited in Trieste' hospitals”. SIGU meeting 2021 (online).
- Tesolin P, Fiorino S, Lenarduzzi S, Cattaruzzi E, Ammar L, Castro V, Orzan E, Granata C, Morgan A, Girotto G. “Pendred syndrome, or not Pendred syndrome? that is the question”. SIGU meeting 2021 (online).
- Morgan A, La Bianca M, Nardone GG, Alessandrini B, Severi G, Graziano C, Gasparini P, Faletra F, Girotto G. “Whole Exome Sequencing Shed the Light on the Possibility of Dual Molecular Diagnoses for Unusual Hearing Loss Cases”. ARO meeting 2022 (online).
- Tesolin P, Fiorino S, Nardone GG, Alessandrini B, Lenarduzzi S, Cattaruzzi E, Ammar L, Dell’Orco D, Granata C, Morgan A, Girotto G. “Molecular Diagnosis for Patients Affected by Pendred Syndrome: A New Genotype-Phenotype Correlation and a Novel Candidate Gene”. ARO meeting 2022 (online).
- Luglio A, Rubinato E, Bonati MT, Faletra F, Feresin A, Morgan A, Morgante G, Musante L, Spedicati B, Girotto G, Gasparini P. “PERCHING syndrome: extremely rare and complicate clinical diagnosis”. ESHG 2022, Vienna, Austria (Poster presentation).
- Alessandrini B, Lenarduzzi S, Dal Ferro M, Paldino A, Sinagra G, Gasparini P, Girotto G. “Cardiomyopathies (CMs): from clinical characterization to Whole Exome Sequencing (WES) analysis in Italian patients”. ESHG 2022, Vienna, Austria (Poster presentation).

- Feresin A, Bonati MT, Faletra F, Girotto G, Luglio A, Morgan A, Morgante G, Rubinato E, Spedicati B, Tesser A, Tommasini A, Gasparini P. "PSMD12 haploinsufficiency is not simply a Neurodevelopmental Disorder". ESHG 2022, Vienna, Austria (Poster presentation).
- Morgan A, Lenarduzzi S, Spedicati B, Tesolin P, Santin A, Rubinato E, Girotto G. "Accurate clinical evaluation and high throughput technologies for the molecular characterization of hereditary hearing loss in a large cohort of Italian patients". ESHG 2022, Vienna, Austria (Poster presentation).
- Tesolin P, Bertinetto F, Sonaglia A, Concas MP, Morgan A, Nardone GG, Zabotti A, Gasparini P, Amoroso A, Quartuccio L, Girotto G. "High throughput genetic characterization of Italian patients affected by multidrug resistant rheumatoid and psoriatic arthritis". ESHG 2022, Vienna, Austria (Poster presentation).
- Nardone GG, Concas MP, Loperfido D, Morgan A, Battaglia Parodi M, Gasparini P, Girotto G. "Dissecting the genetic bases of Color Vision Defects (CVDs) through Genome-Wide Association Study (GWAS) in Silk Road populations". ESHG 2022; Vienna, Austria (Poster presentation).
- Santin A, Tesolin P, Spedicati B, Zito G, Romano F, Morgan A, Concas MP, Ricci G, Girotto G. "Shedding light on Endometriosis (EM) disease: Whole Exome Sequencing (WES) and new genes discovery in a fully clinical characterized Italian cohort." ESHG 2022; Vienna, Austria (Poster presentation).
- Morgante G, Bonati MT, Faletra F, Morgan A, Rubinato E, Feresin A, Luglio A, Spedicati B, Girotto G, Gasparini P. "Whole Exome Sequencing (WES) in unravelling complex cases of bicuspid aortic valve". ESHG 2022, Vienna, Austria (Poster presentation).
- Morgan A, Lenarduzzi S, Spedicati B, Nardone GG, Tesolin P, Santin A, Rubinato E, Girotto G. "Deep phenotyping and high throughput sequencing technologies for the molecular diagnosis of hereditary hearing loss in an Italian cohort of patients". SIGU 2022; Trieste; Italia (Podium presentation C49).
- 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. "Normal Hearing Function (NHF) and Age-Related Hearing Loss (ARHL): new candidate genes identification from Genome-Wide Association Studies (GWAS) in Moli-sani cohort". SIGU 2022; Trieste, Italia (Podium presentation C10).
- Tesolin P, Morgan A, Pianigiani G, Santin A, Bartoccioni P, Nunes Martinez V, Palacin M, Girotto G. "High-throughput screening: identification of molecules able to promote the transcription of *SLC7A8*". SIGU 2022; Trieste, Italia (Podium presentation C34).
- Feresin A, Luglio B, Spedicati B, Morgan A, Bonati MT, Faletra F, Girotto G, Gasparini P, Rubinato E. "Not only a Klippel-Feil, nor a tube defect: a novel Italian family with V239I *VANGL1".* SIGU 2022; Trieste, Italia (Poster presentation P008).
- Crudele F, Tesolin P, Bertinetto F, Sonaglia A, Concas MP, Morgan A, Ferrero N, Nardone GG, Zabotti P, Gasparini P, Amoroso A, Quartuccio L, Girotto G. "High throughput sequencing technologies allow the genetic characterization of multi-drug resistant patients affected by rheumatoid and psoriatic arthritis". SIGU 2022; Trieste, Italia (Poster presentation P308).
- Nardone GG, Concas MP, Santin A, Morgan A, Gasparini P, Battaglia-Parodi M, Girotto G. "Identifying missing pieces in Color Vision Defects (CVDs): a Genome-Wide Association Study (GWAS) in Silk Road populations". SIGU 2022; Trieste, Italia (Poster presentation P385).
- Nardone GG, Crudele F, Santin A, Morgan A, Concas MP, Girotto G. "Whole Genome Sequencing for the molecular diagnosis of Hearing Loss: overcoming the limits of Whole Exome Sequencing". SIGU 2022; Trieste, Italia (Poster presentation P148).
- 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.

Capacità linguistiche

Lingua	Livello parlato	Livello Scritto
Inglese	C2	C2
Francese	A2	A2
Spagnolo	A2	A2
Portoghese	A1	A1