



## EDUCATION AND TRAINING

2009 – 2014 Venafrò, Italy

**HIGH SCHOOL DIPLOMA (CLASSICAL STUDIES)** Liceo Classico Antonio Giordano

**Final Grade:** 96/100

2014 – 2018 Viterbo, Italy

**BACHELOR DEGREE IN BIOTECHNOLOGY (LM 2)** University of Tuscia

**Thesis:** Role of  $\alpha$ -synuclein in amyloid aggregates in Parkinson's Disease

$\alpha$ -synuclein is an intrinsically disordered protein, responsible for the formation of Lewy bodies in Parkinson's Disease and other synucleinopathies. My thesis focused on the chemico-physical properties of this protein, with particular attention to its propensity to misfold and aggregate into  $\beta$ -sheet-rich fibrillar structures that promote the pathological aggregation of  $\alpha$ -synuclein.

**Final grade:** 104/110

**Bachelor degree internship:** From July to November 2017 at the neuropathology laboratory of Istituto Neurologico Mediterraneo- I.R.C.C.S. "Neuromed". Supervisor Professor Felice Giangaspero. Laboratory supervisor Dr Antonella Arcella.

**Competence acquired:**

- PCR
- Immunohistochemistry
- Tissue samples paraffin embedding

2018 – 2021 Trieste, Italy

**MASTER DEGREE IN MEDICAL BIOTECHNOLOGIES (PHARMACEUTICAL CURRICULUM) (LM-9)** University of Trieste

**Thesis:** Role of C1q in Malignant Mesothelioma Microenvironment: a new possible approach for personalized therapy

Malignant Pleural Mesothelioma is an aggressive tumor mainly caused by asbestos exposition. The main first line of treatment consists of cisplatin, alone or in combination with other chemotherapeutics. However, there's no standardized second line of treatment. My thesis focused on how components of Mesothelioma's microenvironment (i.e Hyaluronic Acid, C1q) can alter its sensibility to chemotherapeutics through the evaluation of the expression of Multi-Drug Resistance proteins and the development of a cisplatin-resistant Mesothelioma cell line.

**Final grade:** 110/110

**Master degree internship:** From June 2020 to July 2021 at the Immunology laboratory. Supervisor Professor Roberta Bulla; Laboratory supervisor Dr Andrea Balduit.

**Competence acquired:**

- Isolation of tumour cells from patients' samples
- 2D cultures of tumor cell lines on plastic and on Hyaluronic acid matrices
- Tumour cell lines pharmacoresistance evaluation:
  - in vitro killing tests with several chemotherapeutics
  - Multi-drug resistance proteins functionality evaluation
- Development of cisplatin-resistant tumor cell line
- Flow Cytometry and flow cytometry data analysis through FlowJo and CellQuest
- Immunofluorescence
- RNA extraction
- Real-time and retrotranscriptase PCR
- Western Blot

- Protein 3D structure modeling through Pymol
- Bioinformatic analysis of proteins' binding motifs

06/2024 – 12/2024 Monserrato, Italy

**VISITING PHD STUDENT** National Research Council (CNR)

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**Supervisor:** Professor Giorgia Girotto, Dr. Serena Sanna

**Research Experience:**

- 16S rRNA sequencing data analyses from both Ion Torrent and Illumina MiSeq sequencing platforms, from data quality control to taxonomic classification of bacteria from the vaginal and intestinal microbial communities.
- Application of advanced statistical methodologies for the analyses of microbial communities from healthy women of the "Women4 Health" (W4H) cohort and women affected by endometriosis.

During my PhD, I had the opportunity to spend six months at the National Research Council. In this period, I've worked under the supervision of Dr. Serena Sanna taking part in different scientific activities and projects conducted by her team. Particularly, I've applied the analysis pipeline used to analyse microbial samples from the W4H cohort to a cohort of ~50 women affected by endometriosis, aiming to find any microbial marker of disease. Moreover, I've applied advanced statistical methodologies to investigate relations between bacterial taxa and pain-related symptoms in the endometriosis cohort, aiming to identify pathogenic taxa and assess the potential for microbiome-modulating therapies in pain management.

2022 – CURRENT Trieste, Italy

**PHD IN PERSONALIZED THERAPIES AND INNOVATIVE MEDICINE** University of Trieste

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**Supervisor:** Professor Giorgia Girotto

**Research Experience:**

- Investigation of performances of a total of 20 Structural Variants (SVs) calling algorithms and 10 alignment algorithms for Whole Genome Sequencing (WGS) data, across classical (short-reads) and upcoming (PacBio, Oxford Nanopore long-reads) sequencing technologies.
- Implementation and maintenance of automated analysis workflows in harmony with the FAIR principles on different types of genetic data with the use of dedicated programming languages (Snakemake, Nextflow).
- Implementation of tailored analysis workflows for Oxford Nanopore sequencing data.
- Administration and maintenance of High Performance Computing (HPC) system.
- Collaboration with national (Network of Italian Genomes - NIG) and international (CHARGE, Cohorts for Heart and Aging Research in Genomic Epidemiology; GIANT, Genetic Investigation of Anthropometric Traits; G-EAR, GLGC, Global Lipid Genetic Consortium) consortia serving respectively as a bioinformatician in the production of aggregated genetic data and as a data analyst performing Genome-Wide Association Studies.

Within the scope of my PhD project, I focused on the evaluation of SVs detection from WGS data. SVs are very difficult to detect from WGS data, with several algorithms available yielding very different accuracy performances. Moreover, SVs are often overlooked during WGS data analysis. Hence, SVs could constitute the molecular basis underlying several undiagnosed conditions. In this light, I've conducted an evaluation of several SVs detection algorithms across different sequencing technologies including classical approaches, such as short-read WGS, and new and upcoming technologies such as PacBio and Oxford Nanopore long-read WGS.

Moreover, during my PhD experience, I've been able to work on the implementation and maintenance of several automated analysis workflows, including SNP array quality control, imputation, WGS analysis and SVs analysis. I've also contributed to the establishment of reliable and reusable analysis frameworks for the analysis of Oxford Nanopore sequencing data.

Finally, during this experience, I've also worked to maintain and administer the HPC system used by Medical Genetics department of I.R.C.C.S Burlo Garofolo, especially in the diagnostic of queue administration software. Moreover, I took part in several research projects of the Medical Genetics department of I.R.C.C.S Burlo Garofolo, spanning several research topics such as hearing loss, endometriosis and olfactory dysfunctions, while continuing my data analyst role within international consortia and taking part as a bioinformatician in the NIG consortium.

## ● WORK EXPERIENCE

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**INSTITUTE FOR MATERNAL AND CHILD HEALTH - IRCCS "BURLO GAROFOLO" – TRIESTE, ITALY**

**GENETICS OF SENSES AND RELATED DISEASES - SCHOLARSHIP – 2021 – 2022**

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**Supervisors:** Professor Giorgia Girotto, Dr. Maria Pina Concas

**Research Experience:**

- Led Whole Genome Sequencing (WGS) data analysis, performing secondary analysis using tools such as FastQC, bwa-mem, and GATK for quality control, alignment, and BAM file production.
- Conducted variant calling, identifying Single Nucleotide Variants (SNVs), small insertions/deletions (indels), and Structural Variants (SVs) from short-read WGS data using specialized software (Manta, Delly, Smoove).
- Performed comprehensive VCF file annotation with Variant Effect Predictor (VEP) to characterize genetic variations.
- Developed and implemented tailored algorithms in Bash, Python, and R for tertiary analysis of WGS and SNP array data.
- Executed Genome-Wide Association Studies (GWAS) using specialized software (e.g. R, GEMMA ).

All the acquired competencies and technical skills have been applied to the study of complex traits and diseases. In particular, I conducted a GWAS study to investigate color vision defects in the unique Silk Road cohort. Moreover, I took part as a data analyst in several international consortia (CHARGE, Cohorts for Heart and Aging Research in Genomic Epidemiology; GIANT, Genetic Investigation of Anthropometric Traits; G-EAR). WGS analyses were mainly applied for the study of rare variants in several outbred and isolated populations with a particular focus on the genetics of senses within the specific project named: "Genetics of senses and related diseases".

## **PUBLICATIONS**

### **Genetic variations associated with the soapy flavor perception in Gorgonzola PDO cheese**

Concas M. P., Piochi M., Cabrino G., Colaianni A., Nardone G. G., Gasparini P., Torri L

<https://doi.org/10.1016/j.foodqual.2022.104569>

2022

### **Genome wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss**

Trpchevska N., Freidin M. B., Broer L., Oosterloo B. C., Yao S., Zhou Y., Vona B., Bishop C., Bizaki-Vallaskangas A., Canlon B., Castellana F., Chasman D. I., Cherny S., Christensen K., Concas M. P., Correa A., Elkon R., Metspalu A., Nelis M., Mägi R., Esko T., Mengel-From J., Gao Y., Giersch A. B. S., Girotto G., Gudjonsson A., Gudnason V., Heard-Costa N. L., Hertzano R., Hjelmberg J. v. B., Hjerling-Leffler J., Hoffman H. J., Kaprio J., Kettunen J., Krebs K., Kähler A. K., Lallemand F., Launer L. J., Lee I., Leonard H., Li C., Lowenheim H., Magnusson P. K. E., van Meurs J., Milani L., Morton C. C., Mäkitie A., Nalls M. A., Nardone G. G., Nygaard M., Palviainen T., Pratt S., Quaranta N., Rämö J., Saarentaus E., Sardone R., Satizabal C. L., Schweinfurth J. M., Seshadri S., Shiroma E., Shulman E., Simonsick E., Spankovich C., Tropitzsch A., Lauschke V. M., Sullivan P. F., Goedegebure A., Cederroth C. R., Williams F. M. K., Nagtegaal A. P

doi: 10.1016/j.ajhg.2022.04.010

2022

### **Implicating genes, pleiotropy, and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis**

Kanoni, S., Graham, S. E., Wang, Y., Surakka, I., Ramdas, S., Zhu, X., Clarke, S. L., Bhatti, K. F., Vedantam, S., Winkler, T. W., Locke, A. E., Marouli, E., Zajac, G. J. M., Wu, K. H., Ntalla, I., Hui, Q., Klarin, D., Hilliard, A. T., Wang, Z., Xue, C., ... Peloso, G. M.

<https://doi.org/10.1186/s13059-022-02837-1>

2023

### **Identifying missing pieces in color vision defects: a genome-wide association study in Silk Road populations**

Nardone G. G., Spedicati B., Concas M. P., Santin A., Morgan A., Mazzetto L., Battaglia-Parodi M., Girotto G.

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

2023

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

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

doi: 10.3390/biomedicines11030703

2023

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

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Spedicati B., Morgan A., Pianigiani G., Musante L., Rubinato E., Santin A., Nardone G. G., Faletra F., Girotto G.

doi: 10.3390/genes13112023

2023

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

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Santin A., Spedicati B., Morgan A., Lenarduzzi S., Tesolin P., Nardone G. G., Mazzà D., Di Lorenzo G., Romano F., Buonomo F., Mangogna A., Concas M. P., Zito G., Ricci G., Girotto G

doi: 10.3390/biomedicines11082122

2024

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

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Santin A, Spedicati B, Pecori A, Nardone GG, Concas MP, Piatti G, Menini A, Tirelli G, Boscolo-Rizzo P, Girotto G

doi: 10.3390/life14020219

2024

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

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Young WJ, van der Most PJ, Bartz TM, Bos MM, Biino G, Duong T, Foco L, Lominchar JT, Müller-Nurasyid M, Nardone GG, Pecori A, Ramirez J, Repetto L, Schramm K, Shen X, van Duijvenboden S, van Heemst D, Weiss S, Yao J, Benjamins JW, Alonso A, Spedicati B, Biggs ML, Brody JA, Dörr M, Fuchsberger C, Gögele M, Guo X, Ikram MA, Jukema JW, Käb S, Kanters JK; Lifelines Cohort Study; Lin HJ, Linneberg A, Nauck M, Nolte IM, Pianigiani G, Santin A, Soliman EZ, Tesolin P, Vaccargiu S, Waldenberger M, van der Harst P, Verweij N, Arking DE, Concas MP, De Grandi A, Girotto G, Grarup N, Kavousi M, Mook-Kanamori DO, Navarro P, Orini M, Padmanabhan S, Pattaro C, Peters A, Pirastu M, Pramstaller PP, Heckbert SR, Sinner M, Snieder H, Völker U, Wilson JF, Gauderman WJ, Lambiase PD, Sotoodehnia N, Tinker A, Warren HR, Noordam R, Munroe PB

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

2025

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

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Spedicati B, Pecori A, Concas MP, Santin A, Ruberto R, Nardone GG, D'Alessandro A, Tirelli G, Boscolo-Rizzo P, Girotto G

doi: 10.3390/life15010056

2025

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

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Toffoli M, Campisciano G, Santin A, Pegoraro S, Zito G, Spedicati B, Balduit A, Romano F, Di Lorenzo G, Mangogna A, Tesolin P, Nardone GG, Zanotta N, Sanna S, Crobu F, Kishore U, Ricci G, Bulla R, Girotto G, Agostinis C

doi: 10.1016/j.lfs.2025.123427

2025

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

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Santin A, Pianigiani G, Gialluisi A, Pecori A, Spedicati B, Costanzo S, Persichillo M, Bracone F, Nardone GG, Tesolin P, Lenarduzzi S, Morgan A, De Curtis A, van der Valk W, Rousset F, Roccio M, Locher H, Iacoviello L, Concas MP, Girotto G

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

2025

### [Polygenic prediction of body mass index and obesity through the life course and across ancestries](#)

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Roelof AJ Smit, Kaitlin H Wade, Qin Hui, Joshua D Arias, Xianyong Yin, Malene R Christiansen et al, (including Nardone GG)

doi: 10.1038/s41591-025-03827-z

2025

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

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Spedicati B, Pasquetti D, Santin A, Zampieri S, Morgan A, Lenarduzzi S, Nardone GG, Paccagnella E, Cappellani S, Diplotti L, Pensiero S, Parentin F, Gasparini P, Battaglia Parodi M, Girotto G

doi: 10.3390/biomedicines13081904

2025

### [A Hitchhiker Guide to Structural Variant Calling: A Comprehensive Benchmark Through Different Sequencing Technologies](#)

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Nardone GG, Andrioletti V, Santin A, Morgan A, Spedicati B, Concas MP, Gasparini P, Girotto G, Limongelli I

doi: 10.3390/biomedicines13081949

2025

### [A Complete Picture of the CYP2D6 Heterogeneity in Northeastern Italian Genetic Isolates](#)

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Tesolin P, Nardone GG, Santin A, Pecori A, Ruberto R, Concas MP, Zampieri S, Girotto G

doi: 10.3390/ijms26199445

2025

### [Variability of transposable elements in six genetic isolates from North-Eastern Italy and their relationship with alcohol consumption, tobacco use and BMI](#)

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Modenini G, Mercuri G, Abondio P, Nardone GG, Santin A, Tesolin P, Spedicati B, Pecori A, Pianigiani G, Concas MP, Girotto G, Gasparini P, Boattini A, Mezzavilla M

doi: 10.1186/s12864-025-12225-1

## ● **ORAL PRESENTATIONS TO SCIENTIFIC CONFERENCES**

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2024

### **Human Genome Meeting (HUGO) 2024, Rome (Italy)**

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Selected abstract for oral communication. Title: "Optimizing Structural Variant Calling: towards a robust and reliable detection from Whole Genome Sequencing"

## ● **POSTER PRESENTATIONS TO SCIENTIFIC CONFERENCES**

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2025

### **European Society of Human Genetics (ESHG) 2025, Milan (Italy)**

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Selected abstract for poster presentation. Title: "A Hitchhiker guide to Structural Variant (SV) Calling: a comprehensive benchmark through short-reads (srWGS) and long-reads (lrWGS) sequencing technologies".

2024

### **Italian Society of Human Genetics (SIGU) 2024 , Padua (Italy)**

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Selected abstract for poster presentation. Title: "Structural Variant (SV) calling optimization: towards a robust and reliable detection from short read Whole Genome Sequencing (srWGS) data".

2023

### **American Society of Human Genetics (ASHG) 2023, Washington D.C. (USA)**

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Selected abstract for poster presentation. Title: "Whole Genome Sequencing (WGS) in COVID-19 persistent chemosensory dysfunction: assessing the role of Loss of Function variants (LoFs) in a highly characterized Italian cohort".

2023

### **European Society of Human Genetics (ESHG) 2023, Glasgow (Scotland)**

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Selected abstract for poster presentation. Title: "Whole Genome Sequencing (WGS) for the molecular diagnosis of Hereditary Hearing Loss (HHL): the underestimated role of Structural Variants (SVs)".

2023

### **Italian Society of Human Genetics (SIGU) 2023 , Rimini (Italy)**

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Selected abstracts for poster presentation. Titles:

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

2022

### **European Society of Human Genetics (ESHG) 2022, Vienna (Austria)**

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Selected abstract for poster presentation. Title: "Dissecting the genetic bases of Color Vision Defects (CVDs) through Genome-Wide Association Study (GWAS) in Silk Road populations"

2022

### **Inner Ear Biology (IEB) 2022, Trieste (Italy)**

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Selected abstract for poster presentation. Title: "Overcoming the limits of Whole Exome Sequencing: the application of Whole Genome Sequencing for the molecular diagnosis of Hearing Loss"

2022

### **Italian Society of Human Genetics (SIGU) 2022, Trieste (Italy)**

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Selected abstracts for poster presentation. Titles:

- "Whole Genome Sequencing for the molecular diagnosis of Hearing Loss: overcoming the limits of Whole Exome Sequencing"
- "Identifying missing pieces in Color Vision Defects (CVDs): a Genome-Wide Association Study (GWAS) in Silk Road populations"

## **CONTRIBUTION TO PRESENTATIONS IN SCIENTIFIC CONFERENCES**

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2022

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

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Anna Morgan, Stefania Lenarduzzi, Beatrice Spedicati, Giuseppe Giovanni Nardone, Paola Tesolin, Aurora Santin, Elisa Rubinato, Giorgia Giroto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2022 meeting.

2022

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

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A. Santin, S. Lenarduzzi, G.G. Nardone, B. Spedicati, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Giallusi, L. Iacoviello, P. Gasparini, M.P. Concas, G. Girotto, on behalf of the Moli-sani Investigators

Selected for oral presentation for Italian Society of Human Genetics (SIGU) 2022 meeting.

2022

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

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A. Santin, P. Tesolin, B. Spedicati, G.G. Nardone, G. Zito, F. Romano, G. Di Lorenzo, A. Morgan, M.P. Concas, G. Ricci, G. Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2022 meeting.

2022

### **Unravelling the genetic bases of persistent olfactory dysfunction in COVID-19 patients: the psychophysical and molecular characterisation of a large Italian cohort**

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B. Spedicati, G.G. Nardone, M.P. Concas, F. Crudele, A. Pecori, A. Santin, G. Tirelli, P. Gasparini, A. Morgan, P. Boscolo-Rizzo, G. Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2022 meeting.

2022

### **High throughput sequencing technologies allow the genetic characterization of multi-drug resistant patients affected by rheumatoid and psoriatic arthritis**

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Francesca Crudele, Paola Tesolin, Francesca Bertinetto, Arianna Sonaglia, Maria Pina Concas, Anna Morgan, Norma Ferrero, Giuseppe Giovanni Nardone, Alen Zabotti, Paolo Gasparini, Antonio Amoroso, Luca Quartuccio, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2022 meeting.

2022

### **Molecular Diagnosis for Patients Affected by Pendred Syndrome: a New Genotype-Phenotype Correlation and a Novel Candidate Gene**

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Paola Tesolin, Sofia Fiorino, Giuseppe Giovanni Nardone, Beatrice Alessandrini, Stefania Lenarduzzi, Elisabetta Cattaruzzi, Lydie Ammar, Daniele Dell'Orco, Claudio Granata, Anna Morgan, Giorgia Girotto

Selected for poster presentation for Association for Research in Otolaryngology (ARO) 2022 meeting.

2022

### **Whole Exome Sequencing Shed the Light on the Possibility of Dual Molecular Diagnoses for Unusual Hearing Loss Cases**

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Anna Morgan, Martina La Bianca, Giuseppe Giovanni Nardone, Beatrice Alessandrini, Giulia Severi, Claudio Graziano, Paolo Gasparini, Flavio Faletra, Giorgia Girotto

Selected for poster presentation for Association for Research in Otolaryngology (ARO) 2022 meeting.

2022

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

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Aurora Santin, Stefania Lenarduzzi, Giuseppe Giovanni Nardone, Beatrice Spedicati, Anna Morgan, Mariarosaria Persichillo, Amalia De Curtis, Simona Costanzo, Alessandro Giallusi, Licia Iacoviello, Paolo Gasparini, Maria Pina Concas, Giorgia Girotto

Selected for oral presentation for Inner Ear Biology (IEB) 2022 meeting

2022

### **There's more behind Hereditary Hearing Loss: molecular and phenotypic expansion of PPP1R12A-related disorder**

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Beatrice Spedicati, Anna Morgan, Livia Garavelli, Giuseppe Giovanni Nardone, Giulia Pelliccione, Giulia Pianigiani, Paolo Gasparini, Giorgia Girotto

Selected for oral presentation for Inner Ear Biology (IEB) 2022 meeting

2022

### **Persistent chemosensory dysfunction in COVID-19 patients: a deep dive into the psychophysical and genetic characterisation of an Italian cohort**

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Beatrice Spedicati, Giuseppe Giovanni Nardone, Aurora Santin, Anna Morgan, Maria Pina Concas, Giancarlo Tirelli, Paolo Gasparini, Paolo Boscolo-Rizzo, Giorgia Girotto

Selected for poster presentation for European Society of Human Genetics (ESHG) 2022 meeting

### **High throughput genetic characterization of Italian patients affected by multidrug resistant rheumatoid and psoriatic arthritis**

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Paola Tesolin, Francesca Bertinetto, Arianna Sonaglia, Maria Pina Concas, Anna Morgan, Giuseppe Giovanni Nardone, Alen Zabotti, Paolo Gasparini, Antonio Amoroso,, Luca Quartuccio, Giorgia Girotto

Selected for poster presentation for European Society of Human Genetics (ESHG) 2022 meeting

2023

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

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Aurora Santin, Stefania Lenarduzzi, Giuseppe Giovanni Nardone, Beatrice Spedicati, Anna Morgan, Mariarosaria Persichillo, Amalia De Curtis, Simona Costanzo, Alessandro Gialluisi, Licia Iacoviello, Paolo Gasparini, Maria Pina Concas, Giorgia Girotto

Selected for poster presentation for Inner Ear Biology (IEB) 2023 meeting

2023

### **Unravelling the Genetic Bases of Hearing Loss: a Multistep and Integrative Approach in a Deeply Characterised Italian Cohort**

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Beatrice Spedicati, Aurora Santin, Giuseppe Giovanni Nardone, Stefania Lenarduzzi, Elisa Paccagnella, Elisa Rubinato, Anna Morgan, Giorgia Girotto

Selected for poster presentation for Inner Ear Biology (IEB) 2023 meeting

2023

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

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Anna Morgan, Beatrice Spedicati, Aurora Santin, Giuseppe Giovanni Nardone, Stefania Lenarduzzi, Elisa Paccagnella, Elisa Rubinato, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2023 meeting.

2023

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

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Aurora Santin, Beatrice Spedicati, Anna Morgan, Stefania Lenarduzzi, Paola Tesolin, Giuseppe Giovanni Nardone, Silvia Camarda, Harry Stevens, Daniela Mazzà, Giovanni Di Lorenzo, Federico Romano, Francesca Buonomo, Alessandro Mangogna, Maria Pina Concas, Gabriella Zito, Giuseppe Ricci, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2023 meeting.

2023

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

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Aurora Santin, Stefania Lenarduzzi, Giuseppe Giovanni Nardone, Beatrice Spedicati, Anna Morgan, Mariarosaria Persichillo, Amalia De Curtis, Simona Costanzo, Alessandro Gialluisi, Licia Iacoviello, Paolo Gasparini, Maria Pina Concas, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2023 meeting.

2023

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

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

Selected for poster presentation for American Society of Human Genetics (ASHG) 2023 meeting.

2023

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

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A. Santin, S. Lenarduzzi, G.G. Nardone, B. Spedicati, A. Morgan, M. Persichillo, A. De Curtis, S. Costanzo, A. Gialluisi, L. Iacoviello, P. Gasparini, M.P. Concas, G. Girotto

Selected for poster presentation for European Society of Human Genetics (ESHG) 2023 meeting.

2023

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

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

Selected for poster presentation for European Society of Human Genetics (ESHG) 2023 meeting.

2024

### **Connecting The Dots Of Endometriosis Disease: The Role Of Genetics, Diet, And Microbiome.**

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Aurora Santin, Alessandro Pecori, Beatrice Spedicati, Anna Morgan, Stefania Lenarduzzi, Paola Tesolin, Giuseppe Giovanni Nardone, Silvia Camarda, Harry Stevens, Giulia Pianigiani, Lara Emily Rosso, Elena Vinerbi, Cristina Bon, Daniela Mazzà, Giovanni Di Lorenzo, Federico Romano, Francesca Buonomo, Alessandro Mangogna, Giuseppina Campisciano, Gabriella Zito, Serena Sanna, Maria Pina Concas, Giuseppe Ricci, Giorgia Girotto

Selected for poster presentation for Human Genome Meeting (HUGO) 2024.

2024

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

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Aurora Santin, Stefania Lenarduzzi, Giuseppe Giovanni Nardone, Alessandro Pecori, Beatrice Spedicati, Anna Morgan, Mariarosaria Persichillo, Amalia De Curtis, Simona Costanzo, Alessandro Gialluisi, Licia Iacoviello, Paolo Gasparini, Maria Pina Concas, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2024 meeting.

2024

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

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Aurora Santin, Alessandro Pecori, Beatrice Spedicati, Anna Morgan, Stefania Lenarduzzi, Paola Tesolin, Giuseppe Giovanni Nardone, Silvia Camarda, Harry Stevens, Giulia Pianigiani, Lara Emily Rosso, Elena Vinerbi, Cristina Bon, Daniela Mazzà, Giovanni Di Lorenzo, Federico Romano, Francesca Buonomo, Alessandro Mangogna, Giuseppina Campisciano, Gabriella Zito, Serena Sanna, Maria Pina Concas, Giuseppe Ricci, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2024 meeting.

2024

### **Clinical application of Nanopore sequencing in the molecular diagnosis of genetic disorders**

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Lenarduzzi Stefania, Anna Morgan, Troian Mabel, Stefania Zampieri, Giuseppe Giovanni Nardone, Bon Cristina, Spedicati Beatrice, Gasparini Paolo, Girotto Giorgia

Selected for oral presentation for Italian Society of Human Genetics (SIGU) 2024 meeting

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

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Paola Tesolin, Giuseppe Giovanni Nardone, Alessandro Pecori, Mabel Troian, Aurora Santin, Lara Emily Rosso, Giulia Pianigiani, Beatrice Spedicati, Giorgia Girotto

Selected for poster presentation for Italian Society of Human Genetics (SIGU) 2024 meeting.

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

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Aurora Santin, Beatrice Spedicati, Anna Morgan, Stefania Lenarduzzi, Paola Tesolin, Alessandro Pecori, Giuseppe Giovanni Nardone, Silvia Camarda, Harry Stevens, Giulia Pianigiani, Lara Emily Rosso, Cristina Bon, Daniela Mazzà, Giovanni Di Lorenzo, Federico Romano, Francesca Buonomo, Alessandro Mangogna, Giuseppina Campisciano, Gabriella Zito, Serena Sanna, Maria Pina Concas, Giuseppe Ricci, Giorgia Girotto

Selected for poster presentation for European Society of Human Genetics (ESHG) 2024 meeting.

## ● SEMINARS

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11/04/2022  
**The Sixth Taste**

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Presented by Dr Catherine Graham.

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

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Presented by Prof. V. R. Sutton

09/03/2022  
**Le displasie scheletriche**

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Presented by Prof. Gioacchino Scarano

09/03/2023  
**Il neonato piccolo per l'età gestionale**

---

Presented by Prof. Luigi Memo

20/03/2023  
**Large scale biobank studies as tools to uncover the genetic background of diseases**

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Presented by Prof. Aarno Palotie

18/05/2023  
**From rare diseases to public health genomics**

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Presented by Prof. Borut Peterlin

04/12/2023  
**Why do we have the diseases we have?**

---

Presented by Prof. Han Brunner

04/12/2023  
**Genetic Architecture of Autism Spectrum Disorders**

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Presented by Prof. Alfredo Brusco

04/12/2023

## Genetics and Genomics in Clinical Psychiatry

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Presented by Prof. Massimo Gennarelli

05/02/2024

## Mutazioni rare e alleli comuni: effetti sulla valutazione del rischio genomico in malattie complesse e multifattoriali

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Presented by Professor Giuseppe Novelli (University of Tor Vergata, Rome, Italy).

05/02/2024

## Research and Diagnostics of rare diseases in Czechia

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Presented by Professor Milan Macek (Charles University, Prague, Czech Republic).

05/02/2024

## Drugs repurposing in RASopathies

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Presented by Professor Giovanni Battista Ferrero (University of Turin, Italy).

14/03/2024

## Development and Plasticity of food preferences at the start of life

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Presented by Dr Luc Marlier (French National Centre for Scientific Research, University of Strasbourg).

19/06/2025

## Mitochondrial disease pathogenesis: vascular dysfunction and inflammation

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Presented by Prof. Valerio Carelli

### ● LANGUAGE SKILLS

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Mother tongue(s): **ITALIAN**

Other language(s):

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken production	Spoken interaction	
<b>ENGLISH</b>	B2	B2	B2	B2	B2

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

### ● SKILLS

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#### Genomic software

Manta | delly | bedtools | SAMtools | fastQC | guppy | Lumpy | Variant Effect Predictor (VEP) | smooove | PLINK | minimap2 | sniffles2 | Survivor | plink2 | BCFtools | fastp | BWA-mem

#### Statistical software

GEM | GraphPad Prism | GEMMA | MMAP | R

#### Flow Cytometry software

FlowJo | CellQuest

#### Proteomic software

Pymol

#### Specific databases

ClinVar | PubMed | gnomAD | Protein Data Bank | HGMD | DGV

## **Programming Languages**

Python | Bash | Snakemake | Nextflow

## **Others**

Microsoft Office | Microsoft Power Point | Microsoft Excel | Microsoft Word

## **High Performing Computing Cluster related**

Docker | SLURM | Singularity | Conda | PBSPro | SunGrid Engine

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*Curriculum ai fini della diffusione online*

25/11/2025