

PERSONAL INFORMATION

Francesca D'Avanzo

Date of birth: 29/01/1985

Nationality: Italian

WORK EXPERIENCE

16/05/2022 - present

Research fellow for the European Reference Network for Hereditary Metabolic Disorders (MetabERN)

Azienda Sanitaria Universitaria Friuli Centrale (ASUFC), Via Pozzuolo 330, 33100 Udine, Italy

Main activities: medical writing, content writing for communication channels and educational activities, management and helpdesk support for the CPMS (Clinical Patient Management System) and other IT platforms and databases

01/04/2021 - 31/05/2022

Biomedical Scientist

Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy

Research Project: Biomarkers identification for the diagnosis of CNS involvement in Mucopolysaccharidosis type II: analysis of patients' iPSC-derived neural cells

15/04/2020 – 31/03/2021

Biomedical Scientist

Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy

Research Project: Paediatric neurodegenerative disorders: optimizing nanoparticle-mediated strategy for brain treatment

15/07/2019 – 15/04/2020

Biomedical Scientist

University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health

Research Fellow in the project: Preclinical evaluation of innovative nanoparticles for CNS therapy in the mouse model of Mucopolysaccharidosis type II

11/03/2019 – 10/07/2019

Biomedical Scientist

Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy

Research Project: Biomarkers identification for the diagnosis of CNS involvement in Mucopolysaccharidosis type II: analysis of patients' iPSC-derived neural cells

01/01/2018 – 16/10/2020

Clinical Study Coordinator

c/o U.O.C Clinica Pediatrica – Azienda Ospedaliera di Padova

Study Coordinator of Clinical Studies on Mucopolysaccharidosis: MPSI Registry (Mucopolysaccharidosis type I), Hunter Outcome Survey (Mucopolysaccharidosis type II), MPS VI Clinical Surveillance Program (Mucopolysaccharidosis type VI)

06/06/2017 – 05/12/2018

Biomedical Scientist

Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy

Research Project: Paediatric neurodegenerative disorders: optimizing nanoparticle-mediated strategy for brain treatment

01/04/2017 – 30/04/2017

Biomedical Scientist

Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy

Research Project: Paediatric neurodegenerative disorders: a preclinical therapeutic strategy by nanoparticles delivery and identification of biomarkers of pathogenesis and treatment efficacy for a personalized medicine approach

- 09/05/2016 – 31/12/2016 **Biomedical Scientist**
Pediatric Research Institute, Corso Stati Uniti 4, 35127 Padua, Italy
Research Project: Paediatric neurodegenerative disorders: a preclinical therapeutic strategy by nanoparticles delivery and identification of biomarkers of pathogenesis and treatment efficacy for a personalized medicine approach
- 01/09/2013 – 31/03/2016 **Collaborator in the European Project "Inherited NeuroMetabolic Disease Information Network" (InNerMeD-I-Network, 2012 12 12, Second Health Programme 2008-2013)**
Brains for Brain Foundation Onlus - Via Giustiniani, 3 - Padova
Main activities: design and management of a database on neurometabolic diseases, disease classification, and data entry; member of the editorial board: creation and content management of various communication channels (website, newsletters, publications, social networks and conferences)
- 01/05/2012 – 30/04/2013 **Biomedical Scientist**
University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health
Research fellow in the project: Therapy of Metachromatic Leukodystrophy through Neural Stem Cells
- 01/05/2011 – 31/05/2011 **Biomedical Scientist**
Brains for Brain Foundation Onlus - Via Giustiniani, 3 - Padova
Research Project: Evaluation of the efficacy of Enzymatic Replacement Therapy in paediatric Mucopolysaccharidosis type II patients
- 01/04/2010 – 31/03/2011 **Biomedical Scientist**
University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health
Scholarship fellow in the research project: Evaluation of the Enzymatic Replacement Therapy in paediatric patients with Mucopolysaccharidosis type II. Search for molecular markers of therapeutic efficacy
- 01/01/2010 – 31/03/2010 **Biomedical Scientist**
Brains for Brain Foundation Onlus - Via Giustiniani, 3 - Padova
Research Project: Nanoparticle - mediated therapy in MPSII mouse model
- 01/01/2010 – 31/12/2012 **PhD Student in "Developmental Medicine and Health Planning Sciences. Curriculum: Rare Diseases; Genetics, Biology and Biochemistry"**
University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health
- 15/11/2009 - 10/12/2009 **Biomedical Scientist**
Brains for Brain Foundation Onlus - Via Giustiniani, 3 - Padova
Research Project: Nanoparticle - mediated therapy in MPSII mouse model
- 17/10/2008 – 14/11/2009 **Internship as graduate student**
University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health
- 10/01/2008 - 16/10/2008 **Master's degree internship**
University of Padova - Via 8 Febbraio, 2 – Padova. Laboratory of Diagnosis and Therapy of Lysosomal Disorders - Department of Women's and Children's Health

05/2006 – 09/2006

Bachelor's degree internship

University of Padova - Via 8 Febbraio, 2 – Padova. BioComputing UP Laboratory - Department of Biomedical Sciences

EDUCATION AND TRAINING

12/09/2013	Professional course: "Innovative technologies in molecular medicine"	EQF level: 7
17/04/2013	PhD in "Developmental Medicine and Health Planning Sciences. Curriculum: Rare Diseases; Genetics, Biology and Biochemistry" University of Padova - Via 8 Febbraio, 2 - Padova Thesis title: Analysis of Hunter syndrome by RNA-sequencing	EQF level: 8
16/10/2008	Master Degree in Molecular Biology (110/110) University of Padova - Via 8 Febbraio, 2 - Padova Thesis title: Evaluation of possible therapeutic strategies for the neurological involvement in Mucopolysaccharidosis type II	EQF level: 7
29/09/2006	Bachelor Degree in Molecular Biology (110/110 cum laude) University of Padova - Via 8 Febbraio, 2 - Padova Thesis title: <i>In silico</i> analysis of <i>Drosophila melanogaster</i> gene Timeless	EQF level: 6
2005	Professional course: "Expert in methods and applications of molecular biology techniques" University of Padova - Via 8 Febbraio, 2 - Padova 323-hour course, in the framework of "Programma Operativo Regionale del Fondo Sociale Europeo"	
2003	Scientific High School Diploma (98/100) Liceo Scientifico Statale di Rotonda (PZ)	EQF level: 4
2002	ECDL "European Computer Driving Licence" AICA (Associazione Italiana per l'Informatica e il Calcolo Automatico) 7 modules: Concepts of ICT, Using the Computer and Managing files, Word Processing, Spreadsheets, Databases, Presentations, Web Browsing and Communication	

PERSONAL SKILLS

Mother tongue(s) Italian

Other language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	B2	B2	B2	B2	B2
Esperanto	B2	B2	B2	B2	B2

Levels: A1/A2: Basic user - B1/B2: Independent user - C1/C2 Proficient user

Digital skills

SELF-ASSESSMENT				
Information processing	Communication	Content creation	Safety	Problem solving
Proficient user	Proficient user	Proficient user	Proficient user	Proficient user

Levels: Basic user - Independent user - Proficient user

[Digital competences - Self-assessment grid](#)

- Excellent mastery of office suite tools (word processor, spreadsheet, presentation software, database management), in possession of ECDL European computer driving license
- Basic knowledge of PERL, Matlab and R programming languages
- Database design and management, knowledge of MySQL language
- Familiarity with the main bioinformatic databases of DNA, RNA and protein sequences, protein interactions, gene expression, genetic variants, diseases and gene-phenotype associations, medical information, clinical trials.
- Use of bioinformatics software for the identification of differentially expressed genes, functional analysis of gene lists and gene expression data, clustering, pathway and network analysis, visualization of gene variants, and prediction of their effects.
- Use of image management software
- Use of citation and reference management software
- Website content management
- Management of digital communication channels

ADDITIONAL INFORMATION

Publications

Web of Science ResearcherID H-5081-2016
Scopus Author ID 6505904081
ORCID 0000-0002-2790-6481
H-index 13

Badenetti L, Manzoli R, Trevisan M, D'AVANZO F, Tomanin R, Moro E (2023). A novel CRISPR/Cas9-based iduronate-2-sulfatase (IDS) knockout human neuronal cell line reveals earliest pathological changes. SCIENTIFIC REPORTS;13(1):10289. doi:10.1038/s41598-023-37138-5.

D'AVANZO F, Zanetti A, Dardis A, Scarpa M, Volpi N, Gatto F, Tomanin R (2023). Mucopolysaccharidoses Differential Diagnosis by Mass Spectrometry-Based Analysis of Urine Free Glycosaminoglycans-A Diagnostic Prediction Model. BIOMOLECULES;13(3):532. doi:10.3390/biom13030532

Maccari F, Rigon L, Mantovani V, Galeotti F, Salvalaio M, D'AVANZO F, Zanetti A, Capitani F, Gabrielli O, Tomanin R, Volpi N (2022). Glycosaminoglycan signatures in body fluids of mucopolysaccharidosis type II mouse model under long-term enzyme replacement therapy. JOURNAL OF MOLECULAR MEDICINE (Berlin);100(8):1169-1179. doi: 10.1007/s00109-022-02221-3.

Casamassa A, Zanetti A, Ferrari D, Lombardi I, Galluzzi G, D'AVANZO F, Cipressa G, Bertozzi A, Torrente I, Vescovi AL, Tomanin R, Rosati J (2022). Generation of an induced pluripotent stem cells line, CSSi014-A 9407, carrying the variant c.479C>T in the human iduronate 2-sulfatase (hIDS) gene. STEM CELLS RESEARCH;63:102846. doi: 10.1016/j.scr.2022.102846.

Sestito S, Rinninella G, Rampazzo A, D'AVANZO F, Zampini L, Santoro L, Gabrielli O, Fiumara A, Barone R, Volpi N, Scarpa M, Tomanin R, Concolino D (2022). Cardiac involvement in MPS patients: incidence and response to therapy in an Italian multicentre study. ORPHANET JOURNAL OF RARE DISEASES;17(1):251. doi: 10.1186/s13023-022-02396-5.

D'AVANZO F, Zanetti A, De Filippis C, Tomanin R (2021). Mucopolysaccharidosis Type VI, an Updated Overview of the Disease. INTERNATIONAL JOURNAL OF MOLECULAR SCIENCE;22(24):13456.

doi: 10.3390/ijms222413456.

Zanetti A, D'AVANZO F, AISayed M, Brusius-Facchin AC, Chien YH, Giugliani R, Izzo E, Kasper DC, Lin HY, Lin SP, Pollard L, Singh A, Tonin R, Wood T, Morrone A, Tomanin R (2021). Molecular basis of mucopolysaccharidosis IVA (Morquio A syndrome): A review and classification of GALNS gene variants and reporting of 68 novel variants. *HUMAN MUTATION*;42(11):1384-1398. doi:10.1002/humu.24270.

Zanetti A, D'AVANZO F, Bertoldi L, Zampieri G, Feltrin E, De Pascale F, Rampazzo A, Forzan M, Valle G, Tomanin R (2020). Setup and Validation of a Targeted Next-Generation Sequencing Approach for the Diagnosis of Lysosomal Storage Disorders. *THE JOURNAL OF MOLECULAR DIAGNOSTICS*;22(4):488-502. doi: 10.1016/j.jmoldx.2020.01.010

D'AVANZO F, Rigon L, Zanetti A, Tomanin R (2020). Mucopolysaccharidosis Type II: One Hundred Years of Research, Diagnosis, and Treatment. *INTERNATIONAL JOURNAL OF MOLECULAR SCIENCE*;21(4):1258. doi: 10.3390/ijms21041258.

Rigon L, Salvalaio M, Pederzoli F, Legnini E, Belletti D, D'AVANZO F, De Filippis C, Ruozi B, Marin O, Vandelli MA, Forni F, Scarpa M, Tosi G, Tomanin R (2019). Targeting brain disease in MPS II: in vitro and in vivo preclinical efficacy of g7-PLGA nanoparticles. *INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES*;20(8). pii: E2014. doi: 10.3390/ijms20082014

Zanetti A, D'AVANZO F, Rigon L, Rampazzo A, Concolino D, Barone R, Volpi N, Santoro L, Lualdi S, Bertola F, Scarpa M, Tomanin R (2019). Molecular diagnosis of patients affected by mucopolysaccharidosis: a multicenter study. *EUROPEAN JOURNAL OF PEDIATRICS*; 178(5):739-753 doi: 10.1007/s00431-019-03341-8.

Salvalaio M, Rigon L, D'AVANZO F, Legnini E, Balmaceda Valdez V, Zanetti A, Tomanin R (2017). Targeting Brain Disease in Mucopolysaccharidoses. In: Giovanni Tosi editor. *Nanomedicine and Neuroscience: Advantages, Limitations and Safety Aspects*. Ebook series: *Frontiers in Nanomedicine*. Bentham Science Publishers; 2:156-183

Salvalaio M, D'AVANZO F, Rigon L, Zanetti A, D'Angelo M, Valle G, Scarpa M, Tomanin R (2017). Brain RNA-Seq Profiling of the Mucopolysaccharidosis Type II Mouse Model. *INTERNATIONAL JOURNAL OF MOLECULAR SCIENCES*; 18(5). pii: E1072. doi: 10.3390/ijms18051072
CORRESPONDING AUTHOR. IF2016: 3,226

Bertoldi L, Forcato C, Vitulo N, Birolo G, De Pascale F, Feltrin E, Schiavon R, Anglani F, Negrisolo S, Zanetti A, D'AVANZO F, Tomanin R, Faulkner G, Vezzi A, Valle G (2016). QueryOR: a comprehensive web platform for genetic variant analysis and prioritization. *BMC BIOINFORMATICS*; 18(1):225. doi: 10.1186/s12859-017-1654-4. IF2016: 2,448

Giannuzzi V, Devlieger H, Margari L, Odilind VL, Ragab L, Bellettato CM, D'AVANZO F, Lampe C, Schüttler K, Cassis L, Cortès-Saladelafont E, Garcia-Cazorla A, Barić I, Cvitanović-Šojat L, Fumić K, I Dali C, Bartoloni F, Bonifazi F, Scarpa M, Ceci A (2016). The ethical framework for performing research with rare inherited neurometabolic disease patients. *EUROPEAN JOURNAL OF PEDIATRICS*; 176(3):395-405. doi: 10.1007/s00431-017-2852-9. IF2016: 1,921

Zalfa C, Verpelli C, D'AVANZO F, Tomanin R, Vicidomini C, Cajola L, Manara R, Sala C, Scarpa M, Vescovi AL, De Filippis L. (2016). Glial degeneration with oxidative damage drives neuronal demise in MPSII disease. *CELL DEATH AND DISEASE*; 7:e2331. doi:10.1038/cddis.2016.231. IF2016: 5,965

Salvalaio M, Rigon L, Belletti D, D'AVANZO F, Pederzoli F, Ruozi B, Marin O, Vandelli MA, Forni F, Scarpa M, Tomanin R, Tosi G (2016). Targeted Polymeric Nanoparticles for Brain Delivery of High Molecular Weight Molecules in Lysosomal Storage Disorders. *PLOS ONE*; 11(5):e0156452. doi:10.1371/journal.pone.0156452. IF2016: 2,806

Cassis L, Cortès-Saladelafont E, Molero-Luis M, Yubero D, González MJ, Herrero A, Fons C, Jou C, Sierra C, Castejon Ponce E, Ramos F, Armstrong J, O'Callaghan MM, Casado M, Montero R, Olivas SM, Artuch R, Barić I, Bartoloni F, Bellettato CM, Bonifazi F, Ceci A, Cvitanović-Šojat L, Dali CI, D'AVANZO F, Fumić K, Giannuzzi V, Lampe C, Scarpa M, Cazorla ÁG (2015). Review and evaluation of the methodological quality of the existing guidelines and recommendations for inherited neurometabolic disorders. *ORPHANET JOURNAL OF RARE DISEASES*; 10:164. doi: 10.1186/s13023-015-0376-9. IF2016: 3,507

Rigon L, Salvalaio M, Tosi G, Belletti D, D'AVANZO F, Ruozi B, Vandelli MA, Forni F, Scarpa M and Tomanin R (2014). BBB Crossing in Lysosomal Storage Disorders: a Nanoparticle-Based Approach. Abstracts of Free Communications Accepted for Presentation at the 13th International Symposium on

Mucopolysaccharidoses and Related Diseases, Sauipe, Bahia, Brazil, August 13-17, 2014. JOURNAL OF INBORN ERRORS OF METABOLISM & SCREENING, January 2014; 2, doi:10.1177/2326409814538909. IF2016: 3,226

Tomanin R, Zanetti A, D'AVANZO F, Rampazzo A, Gasparotto N, Parini R, Pascarella A, Concolino D, Procopio E, Fiumara A, Borgo A, Frigo AC and Scarpa M (2014). Clinical efficacy of Enzyme Replacement Therapy in paediatric Hunter patients, an independent study of 3.5 years. ORPHANET JOURNAL OF RARE DISEASES; 9:129. doi:10.1186/s13023-014-0129-1. IF2016: 3,507

Fusar Poli E, Zalfa C, D'AVANZO F, Tomanin R, Carlessi L, Bossi M, Nodari LR, Binda E, Marmioli P, Scarpa M, Delia D, Vescovi AL and De Filippis L (2013). Murine neural stem cells model Hunter disease in vitro: glial cell-mediated neurodegeneration as a possible mechanism involved. CELL DEATH AND DISEASE; 4:e906. doi:10.1038/cddis.2013.430. IF2016: 5,965

Mazzocchi G, Tomanin R, Mazza T, D'AVANZO F, Salvalaio M, Rigon L, Zanetti A, Pazienza V, Francavilla M, Giuliani F, Vinciguerra M and Scarpa M (2013). Circadian transcriptome analysis in human fibroblasts from Hunter syndrome and impact of iduronate-2-sulfatase treatment. BMC MEDICAL GENOMICS; 6:37. doi:10.1186/1755-8794-6-37. IF2016: 2,848

Tomanin R, Bellettato CM, D'AVANZO F, Zanetti A, Ceci A, Begley D, Scarpa M (2013). Personalized Medicine In Rare Paediatric Neurometabolic Diseases. In: Silvia Mandel editor: "Neurodegenerative Diseases: Integrative PPPM Approach as the Medicine of the Future". (book series: Olga Golubnitschaja editor. Advances in Predictive, Preventive and Personalised Medicine). Springer; 2:311-327

Tomanin R, Zanetti A, Zaccariotto E, D'AVANZO F, Bellettato CM and Scarpa M (2012). Gene therapy approaches for Lysosomal Storage Disorders, a good model for the treatment of mendelian diseases. ACTA PAEDIATRICA; 101(7):692-701. doi:10.1111/j.1651-2227.2012.02674.x. IF2016: 2,043

Scarpa M, Zaccariotto E, D'AVANZO F, van Weperen W, Rip J, Gaillard P and Tomanin R (2011). First evidence of efficacy in brain following intravenous injections of Brain-Targeted Liposome-Enzyme in the MPS II mouse model. MOLECULAR GENETICS AND METABOLISM; 102: S40-S40; doi:10.1016/j.ymgme.2010.11.135 – Abstract. IF2016: 3,769

Conferences and Workshops

Participation in more than 40 scientific conferences and workshops in the field of Inherited Metabolic Diseases and Lysosomal Storage Disorders, Genetics, and Bioinformatics, presenting abstracts, posters, or oral presentations.

Prizes and Awards

14-17 September 2017: Travel grant AIMPS (Associazione Italiana Mucopolisaccaridosi e malattie affini) ONLUS for "21st ESGLD (European Study Group on Lysosomal Diseases) Workshop - Lyon, France 14-17 September 2017".

23-26 November 2017: Best Poster at XIX National Congress of the Italian Society of Human Genetics (SIGU) - Torino 23-26 November 2016, with the following poster and related oral presentation: "Enzyme-loaded Nanoparticles for CNS drug delivery in genetic metabolic disorders: application to Mucopolysaccharidosis II".

01 May 2012 - 1-year Fellowship University of Padova - Department of Women's and Children's Health for the project: "Therapy of Metachromatic Leukodystrophy through neural stem cells".

01 April 2010 - 1-year Scholarship University of Padova - Department of Women's and Children's Health with the project: "Evaluation of the Enzymatic Replacement Therapy in paediatric patients with Mucopolysaccharidosis type II. Search for molecular markers of therapeutic efficacy."

10-13 September 2009: Travel grant AIMPS (Associazione Italiana Mucopolisaccaridosi e malattie affini) ONLUS for "17th ESGLD (European Study Group on Lysosomal Diseases) Workshop - Bad Honnef (Germany) 10-13 September 2009"

La sottoscritta Francesca D'Avanzo, a conoscenza di quanto prescritto dall'art. 76 del D.P.R. 28 dicembre 2000 n. 445, sulla responsabilità penale cui può andare incontro in caso di falsità in atti e di dichiarazioni mendaci, nonché di quanto prescritto dall'art. 75 del D.P.R. 28 dicembre 2000 n. 445, sulla decadenza dai benefici eventualmente conseguenti al provvedimento emanato sulla base di dichiarazioni non veritiere, ai sensi e per gli effetti del citato D.P.R. n. 445/2000 e sotto la propria personale responsabilità, dichiara che tutte le informazioni contenute nel presente curriculum vitae sono veritiere.

Padova, 3rd December 2023