

BIOGRAPHICAL SKETCH

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NAME: Trivellin, Giampaolo (giampaolo.trivellin@hunimed.eu)

eRA COMMONS USER NAME (credential, e.g., agency login): TRIVELLING

POSITION TITLE: Assistant Professor (Medical Genetics)

EDUCATION/TRAINING (*Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.*)

INSTITUTION AND LOCATION	DEGREE (if applicable)	Start Date MM/YYYY	Completion Date MM/YYYY	FIELD OF STUDY
University of Padova, Italy	BSc	10/2002	07/2005	Molecular Biology
University of Padova, Italy	MSc	10/2005	07/2007	Sanitary Biology
University of Padova, Italy	Ph.D.	01/2008	04/2011	Clinical Methodology and Endocrinological Sciences
Queen Mary University of London (QMUL)	Postdoctoral Research Assistant	02/2011	02/2013	Genetics of pituitary tumors
National Institutes of Health (NIH)	Postdoctoral Visiting Fellow	03/2013	03/2018	Genomics of pituitary gigantism

A. Personal Statement

My research activity focuses on the study of genetic defects causing tumors in the pituitary gland, specifically those leading to gigantism in children and acromegaly in adults. This line of study originated during my PhD in Padova, Italy, in the laboratory of Prof. Carla Scaroni. It continued through my first post-doctoral experience in Prof. Marta Korbonits's lab at Queen Mary University of London, UK. Subsequently, I joined Dr. Constantine Stratakis's lab at the National Institutes of Health in the USA. In 2014, while in his lab, I identified a new gene—an orphan receptor named *GPR101*—involved in the most extreme cases of infantile-onset pituitary gigantism. Following this discovery, my work focused on identifying GPR101 ligands and understanding how this receptor regulates children's growth, utilizing transgenic zebrafish models and cell lines that I developed.

In 2020, I started a junior investigatorship (Marie Curie fellowship) at the Humanitas Research Hospital in Milan, Italy. In 2021, I received a grant from the Italian Telethon Foundation for research on rare genetic diseases, enabling me to continue my research for the next three years. My current independent research interest is to functionally characterize GPR101 by dissecting the epigenetic mechanisms controlling its transcriptional regulation in normal and tumor pituitary cells. Additionally, I aim to quantify the intracellular effects elicited by newly discovered GPR101 inhibitors. I currently hold the positions of Assistant Professor of Medical Genetics at Humanitas University and of Neuroendocrinology referent in the laboratory of Translational Endocrinology and Metabolism at the Humanitas Research Hospital.

Ongoing and recently completed projects that I would like to highlight include:

- NRRP - Mission 4 "Education and Research", Funding projects presented by young researchers

Project title: Decoding chromatin structure and phenotypic changes in X-linked hypopituitarism using human iPSC-derived 3D pituitary cultures

Duration: 36 months (start date: December 20, 2022), total grant amount: €300,000.00

- Telethon Foundation General Grant 2020

Project number: GGP20130

Project title: Illuminating the biology of the GPR101 receptor: analysis of its transcriptional regulation and validation of new ligands

Duration: 36 months (start date: June 01, 2021), total grant amount: €238,788.00

- Society for Endocrinology (SfE) Equipment Grant – May 2019

Total grant amount: UK £9,100 awarded to purchase vital equipment to establish my own laboratory

- Horizon 2020, Marie Skłodowska Curie Action (MSCA) - Individual Fellowship 2018

Project number: 843843

Project title: Identification and functional validation of novel enhancer sequences involved in pituitary gland development and pathology

Duration: 24 months (start date: March 16, 2020), total grant amount: €183,473.28

- 2016 US ASPIRE Investigator Research Award in Endocrinology (Co-PI)

Project title: Characterization of GPR101-mediated growth regulation and receptor deorphanization

Duration: 24 months (start date: July 01, 2016), total grant amount: US \$75,000

B. Positions, Scientific Appointments and Honors

Positions:

- 03/2018 – 03/2020: Research Fellow, NICHD/NIH, USA (Dr. Constantine Stratakis's lab)
- 03/2020 – 12/2022: Researcher (Marie Curie Fellow), Humanitas Research Hospital, Italy (Prof. Andrea Lania's lab)
- 12/2022 – ongoing: Assistant Professor, Humanitas University. Coordinator of the integrated course in Molecular Biology and Fundamentals of Genetics for students enrolled in the BSc course in Biomedical Laboratory Techniques
- 06/2024 – ongoing: Neuroendocrinology referent, laboratory of Translational Endocrinology and Metabolism, Humanitas Research Hospital, Italy

Scientific Appointments:

- 09/2022 – ongoing: Coordinator of the European Neuroendocrine Association (ENEA) Young Researchers Committee (EYRC)
- 03/2024 – ongoing: European Journal of Endocrinology (EJE) Rising Star Reviewer Board

Honors:

- Best oral presentation at the European Neuroendocrinology Association (ENEA) 2020 virtual meeting
- Winner of the 2016 Presidential Poster Competition at ENDO 2016 meeting, Boston, USA
- 2016 NIH Fellows Award for Research Excellence (FARE) competition, Bethesda, USA
- Outstanding Abstract Award at ENDO 2015 meeting, San Diego, USA
- Winner of the 2015 Presidential Poster Competition at ENDO 2015 meeting, San Diego, USA
- 2012 ENDO ESE International Endocrine Scholars Programme - Travel Bursary
- Association for Multiple Endocrine Neoplasia Disorders (AMEND) Young Investigator's Award at SfE BES 2011 meeting, Birmingham, UK

C. Contributions to Science

1. X-linked acrogigantism (X-LAG) syndrome. I contributed to discover and characterize a new syndrome of early childhood-onset pituitary gigantism caused by microduplications of the *GPR101* gene which disrupt the local chromatin architecture.

- [Duplications disrupt chromatin architecture and rewire GPR101-enhancer communication in X-linked acrogigantism.](#)

Franke M, Daly AF, ..., **Trivellin G**

Am J Hum Genet. 2022 Apr 7;109(4):553-570. doi: 10.1016/j.ajhg.2022.02.002

- [The X-linked acrogigantism-associated gene gpr101 is a regulator of early embryonic development and growth in zebrafish.](#)

Trivellin G, Tirosh A, Hernández-Ramírez LC, et al.

Mol Cell Endocrinol. 2021, 520:111091. doi: 10.1016/j.mce.2020.111091

- [Characterization of GPR101 transcript structure and expression patterns.](#)

Trivellin G, Bjelobaba I, Daly AF, et al.

J Mol Endocrinol. 2016 Aug;57(2):97-111. doi: 10.1530/JME-16-0045

- [Gigantism and acromegaly due to Xq26 microduplications and GPR101 mutation.](#)

Trivellin G, Daly AF, Faucz FR, et al.

N Engl J Med. 2014 Dec 18;371(25):2363-74. doi: 10.1056/NEJMoa1408028

2. Test the therapeutic use of a targeted secretion inhibitor in human pituitary tumors. I assessed the ability of a targeted secretion inhibitor directed against SNARE proteins to inhibit GH secretion *ex vivo*. I also contributed to characterize the expression of SNARE proteins in the tumors.

- [Characterization of SNARE proteins in human pituitary adenomas: targeted secretion inhibitors as a new strategy for the treatment of acromegaly?](#)

Garcia EA, **Trivellin G**, Aflorei ED, et al.

J Clin Endocrinol Metab. 2013 Dec;98(12):E1918-26. doi: 10.1210/jc.2013-2602

3. Identification that loss-of-function variants in the PAM gene are enriched in subjects with pituitary hormones hypersecretion. PAM is a bifunctional enzyme that is essential for the biosynthesis of multiple pituitary and hypothalamic peptide hormones. PAM also affects regulated secretion and is involved in the biogenesis of secretory granules.

- [Germline loss-of-function PAM variants are enriched in subjects with pituitary hypersecretion.](#)

Trivellin G, Daly AF, Hernández-Ramírez LC, et al.

Front Endocrinol (Lausanne). 2023 Jun 14;14:1166076. doi: 10.3389/fendo.2023.1166076

4. Study of AIP and CDKN1B (p27) genes in human pituitary tumors. I contributed to functionally characterize the role of both genes in the development of GH-secreting tumors.

- [Interaction of AIP with protein kinase A \(cAMP-dependent protein kinase\).](#)

Scherthaner-Reiter MH, **Trivellin G**, Stratakis CA.

Hum Mol Genet. 2018 Aug 1;27(15):2604-2613. doi: 10.1093/hmg/ddy166

- [A novel mutation in the upstream open reading frame of the CDKN1B gene causes a MEN4 phenotype.](#)

Occhi G, Regazzo D, **Trivellin G**, et al.

PLoS Genet. 2013 Mar;9(3):e1003350. doi: 10.1371/journal.pgen.1003350

- [MicroRNA miR-107 is overexpressed in pituitary adenomas and inhibits the expression of aryl hydrocarbon receptor-interacting protein in vitro.](#)

Trivellin G, Butz H, Delhove J, et al.

Am J Physiol Endocrinol Metab. 2012 Sep;303(6):E708-19. doi: 10.1152/ajpendo.00546.2011

Summary of scientific production:

- complete list of 68 articles (53 original studies and 15 reviews) published in scientific peer-reviewed journals:

<https://www.ncbi.nlm.nih.gov/myncbi/giampaolo.trivellin.1/bibliography/public/>

Scopus metrics (August 12, 2024): H-index 28, total citations 2785