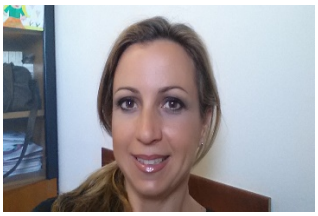
PERSONAL INFORMATION

Annalisa Botta



Tor Vergata University
Dept. of Biomedicine and Prevention
Via Montpellier, 1 – 00133 Rome, Italy
+39 0672596078

botta@med.uniroma2.it

Sex : Female| *Date of birth:* 02/12/1971 | *Nationality:* Italian

Enterprise	University	EPR
☐ Management Level	☐ Full professor	☐ Research Director and 1st level Technologist / First Researcher and 2nd level Technologist
☐ Mid-Management Level	☒ Associate Professor	☐ Level III Researcher and Technologist
☐ Employee / worker level	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator

WORK EXPERIENCE

11/2022 to date	Associate in Medical Genetics, Faculty of Medicine and Surgery, Tor Vergata University of Rome, Italy. Sector: Medical Genetics
2015-2018:	Medical Genetics Counselor, "Tor Vergata" Hospital, Medical Genetics Section, Rome, Italy. Sector: Medical Genetics
2004-10/2022	Assistant Professor in Medical Genetics, Faculty of Medicine and Surgery, Tor Vergata University of Rome, Italy. Sector: Medical Genetics
2001- 2003	Medical Genetics Counselor, "Tor Vergata" Hospital, Medical Genetics Section, Rome, Italy. Sector: Medical Genetics
1999-2001	Telethon Researcher, Medical Genetics Section, Tor Vergata University, Rome, Italy. Sector: Human Genetics
1997-1998	Research Assistant in Medical Genetics, Tor Vergata University, Rome, Italy. Sector: Medical Genetics
1996-1997	Research Assistant in Molecular Biology, Baylor College of Medicine, Houston, TX, USA. Sector: Molecular Biology
1994-1996	Researcher Assistant in Medical Genetics, Tor Vergata University, Rome, Italy. Sector: Medical Genetics

EDUCATION AND TRAINING

Date	From 1999 to 2002	*Replace with EQF (or other) level if relevant*
	Residency in Medical Genetics, MSc	

Title of qualification awarded	Professional qualification for Biologist
Name and type of organization providing education and training	Faculty of Medicine, Sapienza University of Rome, Rome, Italy
Date	1996
Title of qualification awarded	BSc in Biology; degree: 110/110 with highest honors
Name and type of organization providing education and training	Faculty of Science, University of l'Aquila, L'Aquila, Italy
Date	From 1990 to 1995
Title of qualification awarded	High school graduation; degree: 60/60
Name and type of organization providing education and training	Faculty of Science, University of Rome "Tor Vergata", Rome, Itay
Date	From 1992 to July 1997
Title of qualification awarded	
Name and type of organization providing education and training	L.S.S. "Vito Volterra", Rome, Italy

PERSONAL SKILLS

Mother tongue(s)	Italian
Other language(s)	English Level B1 (Understanding, Speaking, Reading)
Job-related skills	Team spirit and good communication skills acquired through the participation to different seminars or meetings. Sense of organization and excellent experience in project management
Digital skills	Good command of Microsoft Office (Word, Excel, PowerPoint), graphic design applications (Photoshop), tools for bioinformatic analyses (Command Console, dChip, NetAffx Analysis Center, DAVID, GSEA, Panther, MirBase, TargetScan), applications for primers design (Primer Express)
Other skills	I am a highly experienced human geneticist with a primary interest in understanding the genetic basis of inherited disorders. I have more than 20 years of experience in human genetics research, more than 15 years leading a productive and active research team collaborating locally and internationally with researchers, clinicians, genetic diagnostic centres and patients' associations. My ongoing work is focused on the study of cis- and trans-acting modifiers of genetic diseases. In this context I am particularly trained in identifying molecular biomarkers (genetic, epigenetic and transcriptional) linked to disease onset and progression. Since 2004, I have been also teaching professor of Medical Genetics courses (both in Italian and English language) in the Faculty of Medicine at Tor Vergata University of Rome.

ADDITIONAL INFORMATION

Publications	Most relevant peer-reviewed publications in the last ten years (from 2013 to 2022)

1. Botta A., Malena A., Tibaldi E., Rocchi L., Loro E., Pena E., Cenci L., Ambrosi E., Bellocchi M.C., Pagano A., Giuseppe Novelli G., Rossi G., Monaco H.L., Gianazza E., Romeo V., Marin O., Brunati A.M., Vergani L. “MBNL142 and MBNL143 gene isoforms, overexpressed in DM1-patient muscle, encode for nuclear proteins interacting with Src-family kinases”. Cell Death Dis. 2013 Aug 15;4(8):e770. doi: 10.1038/cddis.2013.291. PMID: 23949219
2. Botta A., Malena A, Loro E, Del Moro G, Suman M, Pantic B, Szabadkai G, Vergani L. “Altered Ca2+ homeostasis and endoplasmic reticulum stress in myotonic dystrophy type 1 muscle cells”. Genes (Basel). 2013 Jun 4;4(2):275-92. doi: 10.3390/genes4020275. PMID: 24705164. IF:
3. Pantic, B., Borgia, D., Giunco, S., ...Vergani, L., Botta, A.Reliable and versatile immortal muscle cell models from healthy and myotonic dystrophy type 1 primary human myoblasts. Experimental Cell Research, 2016, 342(1), pp. 39–51
4. Perfetti, A., Greco, S., Cardani, R., ...Meola, G., Martelli, F. Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. Scientific Reports, 2016, 6, 38174
5. Botta, A., Rossi, G., Marcaurelio, M., ...Sangiuolo, F., Novelli, G. Identification and characterization of 5′ CCG interruptions in complex DMPK expanded alleles.European Journal of Human Genetics, 2017, 25(2), pp. 257–261
6. Campione, E., Botta, A., Di Prete, M., ...Massa, R., Terracciano, C.Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis.Neuromuscular Disorders, 2017, 27(2), pp. 163–169
7. Ferradini, V., Cassone, M., Nuovo, S., ...Novelli, G., Sangiuolo, F. Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clinica Chimica Acta, 2017, 470, pp. 1–7
8. Santoro, M., Masciullo, M., Silvestri, G., Novelli, G., Botta, A.
9. Santoro, M., Masciullo, M., Silvestri, G., Novelli, G., Botta, A. Myotonic dystrophy type 1: role of CCG, CTC and CGG interruptions within DMPK alleles in the pathogenesis and molecular diagnosis. Clinical Genetics, 2017, 92(4), pp. 355–364
10. Scimeca, M., Centofanti, F., Celi, M., ...Botta, A., Tarantino, U.Vitamin D receptor in muscle atrophy of elderly patients: A key element of osteoporosis-sarcopenia connection.Aging and Disease, 2018, 9(6), pp. 952–964
11. Santoro, M., Fontana, L., Maiorca, F., ...Novelli, G., Botta, A. Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochimica et Biophysica Acta - Molecular Basis of Disease, 2018, 1864(3), pp. 917–924
12. Spitalieri, P., Talarico, R.V., Caioli, S., ...Mango, R., Sangiuolo, F.Modelling the pathogenesis of Myotonic Dystrophy type 1 cardiac phenotype through human iPSC-derived cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2018, 118, pp. 95–109
13. Spitalieri, P., Talarico, R.V., Murdocca, M., ...Sangiuolo, F., Botta, A. Generation and neuronal differentiation of HiPSCs from patients with myotonic dystrophy type 2. Frontiers in Physiology, 2018, 9, 967
14. Binda, A., Renna, L.V., Bosè, F., ...Meola, G., Cardani, R. SCN4A as modifier gene in patients with myotonic dystrophy type 2. Scientific Reports, 2018, 8(1), 11058
15. Bosè, F., Renna, L.V., Fossati, B., ...Meola, G., Cardani, R. TNNT2 missplicing in skeletal muscle as a cardiac biomarker in myotonic dystrophy type 1 but not in myotonic dystrophy type 2.Frontiers in Neurology, 2019, 10(SEP), 992
16. Fontana, L., Santoro, M., D'Apice, M.R., ...Dosa, L., Botta, A. Identification, molecular characterization and segregation analysis of a variant DMPK pre-mutation allele in a three-generation Italian family. Acta Myologica, 2020, 39(1), pp. 13–18
17. D'Apice, M.R., De Dominicis, A., Murdocca, M., ...Federici, M., Novelli, G.Cutaneous and metabolic defects associated with nuclear abnormalities in a transgenic mouse model expressing R527H lamin A mutation causing mandibuloacral dysplasia type A (MADA) syndrome.Acta Myologica, 2020, 39(4), pp. 320–335
18. Centofanti, F., Santoro, M., Marini, M., ...Tarantino, U., Botta, A.Identification of aberrantly-expressed long non-coding rnas in osteoblastic cells from osteoporotic patients.Biomedicines, 2020, 8(3), 65
19. Visconti, V.V., Fittipaldi, S., Ciuffi, S., ...Botta, A., Tarantino, U. Circulating long non-coding rna gas5 is overexpressed in serum from osteoporotic patients and is associated with increased risk of bone fragility. International Journal of Molecular Sciences, 2020, 21(18), pp. 1–12, 6930
20. Nappo, S., Mannucci, L., Novelli, G., ...D'Apice, M.R., Botta, A.Carrier frequency of CFTR variants in the non-Caucasian populations by genome aggregation database (gnomAD)-based analysis.Annals of Human Genetics, 2020, 84(6), pp. 463–468

21. Fittipaldi, S., Visconti, V.V., Tarantino, U., Novelli, G., Botta, A. Genetic variability in noncoding RNAs: Involvement of miRNAs and long noncoding RNAs in osteoporosis pathogenesis.Epigenomics, 2020, 12(22), pp. 2035–2049
22. Botta, A., Visconti, V.V., Fontana, L., ...D'Apice, M.R., Novelli, G. A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. Frontiers in Genetics, 2021, 12, 668094
23. Grosso, V., Marcolungo, L., Maestri, S., ...Delledonne, M., Rossato, M.Characterization of FMR1 Repeat Expansion and Intragenic Variants by Indirect Sequence Capture. Frontiers in Genetics, 2021, 12, 743230
24. Tarantino U, Greggi C, Cariati I, Visconti VV, Gasparini M, Cateni M, Gasbarra E, Botta A, Salustri A, Scimeca M. "The Role of PTX3 in Mineralization Processes and Aging-Related Bone Diseases". Front Immunol. 2021 Jan 29;11:622772. doi: 10.3389/fimmu.2020.622772. eCollection 2020. PMID: 33584725.
25. Spitalieri P, Centofanti F, Murdocca M, Scioli MG, Latini A, Di Cesare S, Citro G, Rossi A, Orlandi A, Miersch S, Sidhu SS, Pandolfi PP, Botta A, Sangiuolo F and Novelli G. "Two Different Therapeutic Approaches for SARS-CoV-2 in hiPSCs-Derived Lung Organoids". Cell. 2022 April 11,1235 doi: 10.3390/cells11071235.
26. Alfano M, De Antoni L, Centofanti F, Visconti VV, Maestri S, Degli Esposti C, Massa R, D'Apice MR, Novelli G, Delledonne M, Botta A, Rossato M. Characterization of full-length *CNBP* expanded alleles in myotonic dystrophy type 2 patients by Cas9-mediated enrichment and nanopore sequencing. Elife. 2022 Aug 26;11:e80229. doi: 10.7554/eLife.80229. PMID: 36018009.
27. Visconti VV, Centofanti F, Fittipaldi S, Macrì E, Novelli G, Botta A. Epigenetics of Myotonic Dystrophies: A Minireview. Int J Mol Sci. 2021 Nov 22;22(22):12594. doi: 10.3390/ijms222212594. PMID: 34830473; PMCID: PMC8623789.
28. Visconti VV, Greggi C, Cariati I, Gasperini B, Mastrogregori A, Botta A, Tarantino U. Deregulated Clusterin as a Marker of Bone Fragility: New Insights into the Pathophysiology of Osteoporosis. Genes (Basel). 2022 Apr 7;13(4):652. doi: 10.3390/genes13040652. PMID: 35456459; PMCID: PMC9024451.
29. Ciuffi S, Marini F, Fossi C, Donati S, Giusti F, Botta A, Masi L, Isaia G, Marcocci C, Migliaccio S, Minisola S, Nuti R, Tarantino U, Iantomasi T, Brandi ML. Circulating MicroRNAs as Biomarkers of Osteoporosis and Fragility Fractures. J Clin Endocrinol Metab. 2022 Jul 14;107(8):2267-2285. doi: 10.1210/clinem/dgac293. PMID: 35532548.
30. Ceccarini MR, Fittipaldi S, Ciccacci C, Granese E, Centofanti F, Dalla Ragione L, Bertelli M, Beccari T, Botta A. Association Between DRD2 and DRD4 Polymorphisms and Eating Disorders in an Italian Population. Front Nutr. 2022 Mar 14;9:838177. doi: 10.3389/fnut.2022.838177. PMID: 35369087; PMCID: PMC8964431.

Complete List of Published Work in MyBibliography:

https://www.scopus.com/authid/detail.uri?authorId=7005993766

Number of total publications (Scopus, updated to 2022): 93

Total number of citations (Scopus, updated to 2022): 2157

H-Index (Scopus, updated to 2022) : 26