



Concetta Scimone

Nationality: Italian

Date of birth: 20/05/1986

Gender: Female

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📍 **Address:** Department of Biomedical and Dental Science and of Morphological and Functional Imaging University of Messina, Via Consolare Valeria 1, 98125 Messina (Italy)

WORK EXPERIENCE

Researcher (RTD-A)

University of Messina [31/12/2021 – Current]

City: Messina

Country: Italy

Researcher (RTD-A) at the Department of Biomedical and Dental Science and of Morphological and Functional Imaging, University of Messina. Discipline: Applied Biology, SSD: BIO/13.

Peer Reviewer (PR) for the Qatar National Research Fund (QNRF)

Qatar National Research Fund (QNRF) [04/2021 – Current]

Country: Qatar

Reviewer Board member for the Qatar National Research Fund (QNRF). The QNRF supports both public and private research activities in the field of life science, energy and development, informatics and social science. Reviewer activity regards projects related to translational research, genomics and personalized medicine, for the National Priorities Research Program (NPRP), the Early Career Researcher Award (ECRA) and the Postdoctoral Research Award (PDRA) research programs.

Research fellow

University of Messina Messina [01/03/2021 – 01/12/2021]

Country: Italy

Research fellow at the Department of Biomedical and Dental Science and of Morphological and Functional Imaging, University of Messina. Research field: "Genetic rare disease: Trimethylaminuria".

Founding partner of the innovative StartUp "D.A.I.R."

[12/2020]

Country: Italy

Founding partner of the innovative StartUp "D.A.I.R." (Data Analysis for Integration in Research) - <https://dair-me.com/>

Professor

United Campus of Malta [11/2020 – 02/2021]

City: Messina

Country: Italy

Neurophysiology professor at the "Physiotherapy" bachelor degree, United Campus of Malta

Principal Investigator - Fellowship

Ordine Nazionale dei Biologi [30/01/2020 – 30/01/2021]

Country: Italy

Principal investigator of the project "A picture of CCM tissue expression by transcriptome analysis: any link with somatic mutations?"

Researcher

Istituto EuroMediterraneo di Scienza e Tecnologia [01/2016 – Current]

City: Palermo

Country: Italy

Researcher at the Istituto EuroMediterraneo di Scienza e Tecnologia (I.E.Me.S.T.), department of "Biomolecula Strategies, Genetics and Avantguard Theraphies.

Honorary fellow

Università degli Studi di Messina [03/2014 – Current]

City: Messina

Country: Italy

Honorary fellow for the discipline Applied Biology, bachelor degree "Medicine and Surgery", Università degli Studi di Messina.

Master professor

Istituto EuroMediterraneo di Scienza e Tecnologia [04/2019 – 11/2019]

City: Palermo

Country: Italy

Teacher of the Master in the field of "Bioinformatics and applied science stistics", organized at the Istituto EuroMediterraneo di Scienza e Tecnologia (I.E.ME.S.T.), Palermo, Italy

Visiting Researcher

Centro Scienze dell'Invecchiamento (Ce.S.I.) - Università degli Studi Gabriele D'Annunzio [04/02/2019 – 07/02/2019]

City: Chieti

Country: Italy

Visiting researcher at the Ce.S.I. (Centro Scienze dell'Invecchiamento), University Gabriele D'Annunzio Chieti-Pescara.

Researcher

Istituto EuroMediterraneo di Scienza e Tecnologia [25/07/2018 – 31/10/2018]

City: Palermo

Country: Italy

Researcher at the l'Istituto I.E.ME.S.T. in the field of the research project "T-MoDiaK (TMAUMolecularDiagnostic Kit)" – PO FESR 2014-2020 Linea di intervento 1.1.2 codice CUP G69J18000460008 – l'attività di: Fase 1 "Progettazione del kit diagnostico", for the development of test for the molecular diagnosis of the trimethylaminuria

Volunteer researcher

University of Messina [01/2015 – 29/01/2019]

City: Messina

Country: Italy

Volunteer researcher at the "Molecular Genetics" laboratory, Department of Biomedical and Dental Science and of Morphological and Functional Imaging, University of Messina.

Professor

University of Messina [01/2016 – 09/2019]

City: Messina

Country: Italy

Professor of "Biology and Genetics" at the bachelor degree "Exercise Science, Sport and Health", University of Messina.

Proferssor

University of Messina [03/2016 – 15/09/2017]

City: Messina

Country: Italy

Professor of "Genetic improvement of animal species" at the bachelor degree "Biotechnology" University of Messina.

INCARICHI EDITORIALI

Guest editor

[09/2021 – Current]

Guest editor of the Special Issue "New Insights on Cellular Biology of Retinal Degenerations" of the "Life" journal (MDPI, Impact Factor 3.817).

https://www.mdpi.com/journal/life/special_issues/Retinal_Degenerations

Guest editor

[09/2020 – Current]

Topic Editor and Guest Editor of the Special Issue "Genetics and Epigenetic Mechanisms of the Neurovascular Unit" of the "Life" journal (MDPI, Impact Factor 3.817).

https://www.mdpi.com/journal/life/special_issues/Genetics_Epigenetic_Mechanisms

Co-autore di libro

[11/2020 – Current]

Co-author of the book: "Advances in Bioinformatics and Statistics" – Volume 1. Bentham Books

ISBN: 978-981-14-8178-9 (Print)

ISBN: 978-981-14-8180-2 (Online)

Year: 2020

DOI:10.2174/97898114818021200101

EDUCATION AND TRAINING

Medical genetics student

University of Messina [11/2018 – Current]

Address: Via Consolare Valeria 1, 98125 Messina (Italy)

Professional qualification

University of Messina [11/2016]

Address: Viale Ferdinando Stagno d'Alcontres 31, 98166 Messina (Italy)

Professional qualification as "Senior Biologist".

High formation course

University of Messina [01/2014 – 04/2015]

Address: Polo Universitario SS Annunziata , 98168 Messina (Italy)

Researcher in "Innovative systems for quality and safety management of animal and vegetal food". "PAN-LAB Project-PON a3_00166 - potenziamento delle strutture e delle dotazioni scientifiche e tecnologiche", program, University of Messina.

PhD in "Biology and cell biology"

University of Messina [04/2014]

Address: Viale Ferdinando Stagno D' Alcontres 31, 98166 Messina (Italy)

Thesis: Basi molecolari delle Malformazioni Cavernose Cerebrali: forme familiari versus forme sporadiche

Master Degree "Biotechnology for Health"

University of Messina [23/07/2010]

Address: Via Consolare Valeria 1, 98125 Messina (Italy)

Final grade : Cum laude

Thesis: Analisi molecolare delle regioni promotrici dei geni CCM

Bechelor Degree "Biotechnology"

University of Messina [08/10/2008]

Address: Via Consolare Valeria 1, 98125 Messina (Italy)

Final grade : Cum laude

Thesis: Analisi mutazionale dei geni coinvolti nello sviluppo delle malformazioni cavernose cerebrali

LANGUAGE SKILLS

Mother tongue(s): **Italian**

Other language(s):

English

LISTENING B2 READING C1 WRITING C1

SPOKEN PRODUCTION B2 SPOKEN INTERACTION B2

DIGITAL SKILLS

Microsoft

Microsoft Word / Microsoft Powerpoint / Microsoft Teams / Microsoft Excel / Microsoft Office Publisher / Microsoft Access / Skype / Outlook

Social

Instagram / Facebook / Twitter / LinkedIn

Online meeting platforms

Zoom / GoToMeeting

Bioinformatic tools and databases

UCSC Genome Browser / Ensembl genome browser / Database biologici online (NCBI EBI NIG) / EMBL / ToppGene / Rfam / GnomAD / Utilizzo di tools per marcatori funzionali (PROSITE, InterPro, EML, Pfam, SignalIP, TMHMM, JPRED) / GeneOntology database / Principali banche dati (PubMed, GeneCards, The Human Protein Atlas) / UniProt / Network analysis Cytoscape / Software per l'analisi dei dati di laboratorio: Galaxy, Pavis, David, IGV, The Meme Suit / Software online per l'analisi di sequenze di DNA e proteine (UCSC Ensembl BLAST ClustalOmega)

AFFILIAZIONE SOCIETÀ SCIENTIFICHE

A.I.B.G. member

[10/2012 – Current]

Member of the "A.I.B.G." - Associazione Italiana di Biologia e Genetica Generale e Molecolare

S.I.G.U. member

[2020]

Member of the "S.I.G.U." – Società Italiana di Genetica Umana

S.I.B.S. member

[04/2021 – Current]

Member of the "S.I.B.S." - Società Italiana di Biologia Sperimentale

E.V.B.O. member

[26/01/2022 – Current]

Member of the "E.V.B.O." - European Vascular Biology Organization

HONOURS AND AWARDS

VEBLEO Fellow

VEBLEO [07/10/2021]

VEBLEO Fellow winner in the context of the "Webinar on Genetics and Molecular Biology" with the Keynote Talk "Molecular signaling in sporadic brain arteriovenous malformation" (September 24-27, 2021).

AUTORIZZAZIONE AL TRATTAMENTO DEI DATI PERSONALI AI SENSI DEL D. LGS. 196/2003 E SS.MM.II.

Autorizzazione al trattamento dei dati personali

Autorizzo l'Università di Messina al trattamento dei dati personali contenuti nel presente curriculum vitae per le finalità connesse con la mia istanza di adesione alla procedura di valutazione in base all'art. 13 del D. Lgs. 196/2003 e all'art. 13 del Regolamento UE 2016/679 relativo alla protezione delle persone fisiche con riguardo al trattamento dei dati personali.

INDICIZZAZIONI ACCADEMICHE

Scopus H-index:18

Co-author of 41 scientific papers published on impacted international journals.

PUBLICATIONS

Evidences of PIEZO1 involvement in cerebral cavernous malformation pathogenesis

[2022]

<https://pubmed.ncbi.nlm.nih.gov/35176291/>

Scimone C, Donato L, Alibrandi S, D'Angelo R, Sidoti A. Evidences of PIEZO1 involvement in cerebral cavernous malformation pathogenesis. *Microvasc Res.* 2022; 141:104342. doi: 10.1016/j.mvr.2022.104342.

Adaptive modelling of mutated fmo3 enzyme could unveil unexplored scenarios linking variant haplotypes to tmau phenotypes

[2021]

<https://www.mdpi.com/1420-3049/26/22/7045>

Alibrandi S, Nicita F, Donato L, Scimone C, Rinaldi C, D'Angelo R, Sidoti A. Adaptive Modelling of Mutated FMO3 Enzyme Could Unveil Unexplored Scenarios Linking Variant Haplotypes to TMAU Phenotypes. *Molecules.* 2021 Nov 22;26(22):7045. doi: 10.3390/molecules26227045. PMID: 34834137; PMCID: PMC8618768.

Investigating the role of imprinted genes in pediatric sporadic brain arteriovenous malformations

[2022]

Scimone C, Donato L, Sidoti A. Investigating the role of imprinted genes in pediatric sporadic brain arteriovenous malformations. *Neural Regen Res.* In press

Oxidative stress and the neurovascular unit

[2021]

<https://www.mdpi.com/2075-1729/11/8/767>

Rinaldi C, Donato L, Alibrandi S, Scimone C*, D'Angelo R, Sidoti A. Oxidative stress and the neurovascular unit. *Life.* 2021; 11(8):767

New evaluation methods of read mapping by 17 aligners on simulated and empirical NGS data: an updated comparison of DNA- and RNA-Seq data from Illumina and Ion Torrent technologies.

[2021]

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8208613/>

Donato L, [Scimone C](#), Rinaldi C, D'Angelo R, Sidoti A. New evaluation methods of read mapping by 17 aligners on simulated and empirical NGS data: an updated comparison of DNA- and RNA-Seq data from Illumina and Ion Torrent technologies. *Neural Comput Appl*. 2021; Jun 16:1-24. doi: 10.1007/s00521-021-06188-z.

N-retinylidene-N-retinylethanolamine adduct induces expression of chronic inflammation cytokines in retinal pigment epithelium cells

[2021]

<https://www.sciencedirect.com/science/article/pii/S0014483521002074?via=ihub>

[Scimone C](#), Donato L, Alibrandi S, Vadalà M, Giglia G, Sidoti A, D'Angelo R. N-retinylidene-N-retinylethanolamine adduct induces expression of chronic inflammation cytokines in retinal pigment epithelium cells. *Exp Eye Res*. 2021; 209:108641. doi: 10.1016/j.exer.2021.108641.

Impairments of photoreceptor outer segments renewal and phototransduction due to a peripherin rare haplotype variant: Insights from molecular modeling

[2021]

<https://www.mdpi.com/1422-0067/22/7/3484>

Donato L, Abdalla EM, [Scimone C*](#), Alibrandi S, Rinaldi C, Nabil KM, D'Angelo R, Sidoti A. Impairments of Photoreceptor Outer Segments Renewal and Phototransduction Due to a Peripherin Rare Haplotype Variant: Insights from Molecular Modeling. *Int J Mol Sci*. 2021 Mar 27;22(7):3484.

Antiretroviral treatment leading to secondary trimethylaminuria: Genetic associations and successful management with riboflavin

[2021]

<https://onlinelibrary.wiley.com/doi/10.1111/jcpt.13315>

[Scimone C](#), Alibrandi S, Donato L, Giofrè SV, Rao G, Sidoti A, D'Angelo R. Antiretroviral treatment leading to secondary trimethylaminuria: Genetic associations and successful management with riboflavin. *J Clin Pharm Ther*. 2021 Apr;46(2):304-309.

Gut-brain axis cross-talk and limbic disorders as biological basis of secondary tmau

[2021]

<https://www.mdpi.com/2075-4426/11/2/87>

Donato L, Alibrandi S, [Scimone C*](#), Castagnetti A, Rao G, Sidoti A, D'Angelo R. Gut-Brain Axis Cross-Talk and Limbic Disorders as Biological Basis of Secondary TMAU. *J. Pers. Med*. 2021; 11(2):87.

New omics-derived perspectives on retinal dystrophies: Could ion channels-encoding or related genes act as modifier of pathological phenotype?

[2021]

<https://www.mdpi.com/1422-0067/22/1/70>

Donato L, [Scimone C*](#), Alibrandi S, Mohamed Abdalla E, Mahmoud Nabil K, D'Angelo R, Sidoti A. New Omics—Derived Perspectives on Retinal Dystrophies: Could Ion Channels-Encoding or Related Genes Act as Modifier of Pathological Phenotype? *Int. J. Mol. Sci*. 2021; 22(1):1–22.

Transcriptome analysis provides new molecular signatures in sporadic Cerebral Cavernous Malformation endothelial cells

[2020]

<https://www.sciencedirect.com/science/article/pii/S0925443920303045?via=ihub>

Scimone C, Donato L, Alibrandi S, Esposito T, Alafaci C, D'Angelo R, Sidoti A. Transcriptome analysis provides new molecular signatures in sporadic Cerebral Cavernous Malformation endothelial cells. *Biochim Biophys Acta Mol Basis Dis.* 2020 Dec 1;1866(12):165956. doi: 10.1016/j.bbadis.2020.165956. Epub 2020 Aug 30. PMID: 32877751.

Expression of Pro-Angiogenic Markers Is Enhanced by Blue Light in Human RPE Cells

[2020]

<https://www.mdpi.com/2076-3921/9/11/1154>

Scimone C, Alibrandi A, Scalinci SZ, Trovato Battagliola E, D'Angelo R, Sidoti A, Donato L. Expression of Pro-Angiogenic Markers Is Enhanced by Blue Light in Human RPE Cells. *Antioxidants.* 2020; 9(11):1–17.

Possible A2E mutagenic effects on RPE mitochondrial DNA from innovative RNA-seq bioinformatics pipeline

[2020]

https://www.mdpi.com/2076-3921/9/11/1158?type=check_update&version=3

Donato L, Scimone C*, Alibrandi S, Pitruzzella A, Scalia F, D'Angelo R, Sidoti A. Possible A2E Mutagenic Effects on RPE Mitochondrial DNA from Innovative RNA-Seq Bioinformatics Pipeline. *Antioxidants.* 2020; 9(11):1–24.

Germline mutation enrichment in pathways controlling endothelial cells homeostasis in patients with

[2020]

<https://www.mdpi.com/1422-0067/21/12/4321>

Scimone C, Granata F, Longo M, Mormina E, Turiaco C, Caragliano AA, Donato L*, Sidoti A*, D'Angelo R. Germline mutation enrichment in pathways controlling endothelial cells homeostasis in patients with brain arteriovenous malformation: implication for molecular diagnosis. *Int J Mol Sci.* 2020; 21(12):43212

Discovery of GLO1 new related genes and pathways by RNA-Seq on A2E-stressed retinal epithelial cell

[2020]

<https://www.mdpi.com/2076-3921/9/5/416>

Donato L, Scimone C, Alibrandi S, Nicocia G, Rinaldi C, Sidoti A, D'Angelo R. Discovery of GLO1 new related genes and pathways by RNA-Seq on A2E-stressed retinal epithelial cells could improve knowledge on retinitis pigmentosa. *Antioxidants (Basel).* 2020; 9(5):416

Effects of A2E-induced oxidative stress on retinal epithelial cells: new insights on retinitis

[2020]

<https://www.mdpi.com/2076-3921/9/4/307>

Donato L, D'Angelo R, Alibrandi S, Rinaldi C, Sidoti A, Scimone C. Effects of A2E-induced oxidative stress on retinal epithelial cells: new insights on retinitis pigmentosa development. *Antioxidants (Basel).* 2020; 9(4):307.

Transcriptome analyses of lncRNAs in A2E-stressed retinal epithelial cells unveil innovative links

[2020]

<https://www.mdpi.com/2076-3921/9/4/318>

Donato L, Scimone C, Alibrandi S, Rinaldi C, Sidoti A, D'Angelo R. Transcriptome analyses of lncRNAs in A2E-stressed retinal epithelial cells unveil innovative links between metabolic impairments related to oxidative stress and retinitis pigmentosa. *Antioxidants (Basel).* 2020; 9(4):318.

High-Throughput Sequencing to detect novel Likely Gene-Disrupting variants in pathogenesis of

[2020]

<https://www.frontiersin.org/articles/10.3389/fgene.2020.00146/full>

Scimone C, Donato L, Alafaci C, Granata F, Rinaldi C, Longo M, D'Angelo R, Sidoti A. High-Throughput Sequencing to detect novel Likely Gene-Disrupting variants in pathogenesis of sporadic brain arteriovenous malformations. *Front Genet.* 2020; 11:146.

Variants of the molecular chaperone HSPA8 and HSPA1A genes in trimethylaminuria: a pilot study

[2020]

Scimone C, Alibrandi S, Donato L, Esposito T, Sidoti A, D'Angelo R. Variants of the molecular chaperone HSPA8 and HSPA1A genes in trimethylaminuria: a pilot study. *EMBJ* 2020, 15 (38) 157-160.

Novel Insights into RPGR Exon ORF15: Could G-Quadruplex Folding Lead to Challenging Sequencing?

[2019]

<http://savvysciencepublisher.com/journal-ocular-diseases-therapeutics-volume-7-open-access/>

Donato L, Scimone C, Rinaldi C, D'Angelo R, Sidoti A. Novel Insights into RPGR Exon ORF15: Could G-Quadruplex Folding Lead to Challenging Sequencing? *Journal of Ocular Diseases and Therapeutics.* 2019; 7:1-11.

Aged fingerprints for DNA profile: First report of successful typing

[2019]

<https://pubmed.ncbi.nlm.nih.gov/31394460/>

Romano CG, Mangiaracina R, Donato L, D'Angelo R, Scimone C*, Sidoti A. Aged fingerprints for DNA profile: First report of successful typing. *Forensic Sci Int.* 2019; 302,109905.

Vis-à-vis: a focus on genetic features of cerebral cavernous malformations and brain arteriovenous

[2019]

<https://link.springer.com/article/10.1007/s10072-018-3674-x>

Scimone C, Donato L, Marino S, Alafaci C, D'Angelo R, Sidoti A. Vis-à-vis: a focus on genetic features of cerebral cavernous malformations and brain arteriovenous malformations pathogenesis. *Neurol Sci.* 2019 Feb;40(2): 243-251.

Association between three polymorphisms in RP1 hotspot region and risk of retinitis pigmentosa in

[2019]

http://www.embj.org/wp-content/uploads/2019/11/Donato_30.pdf

Donato L, Scimone C, Rinaldi C, D'Angelo R, and Sidoti A. Association between three polymorphisms in RP1 hotspot region and risk of retinitis pigmentosa in Italian patients: a pilot study. *EuroMediterranean Biomedical Journal.* 2019; 14(30):130-133.

GLO1 gene polymorphisms and their association with retinitis pigmentosa: a case-control study in a

[2018]

<https://link.springer.com/article/10.1007/s11033-018-4295-4>

Donato L, Scimone C, Nicocia G, Denaro L, Robledo R, Sidoti A, D'Angelo R. GLO1 gene polymorphisms and their association with retinitis pigmentosa: a case-control study in a Sicilian population. *Mol Biol Rep.* 2018; 45(5): 1349-1355.

Stargardt Phenotype Associated With Two ELOVL4 Promoter Variants and ELOVL4 Downregulation: New

[2018]

<https://iovs.arvojournals.org/article.aspx?articleid=2672672>

Donato L, Scimone C, Rinaldi C, Aragona P, Briuglia S, D'Ascola A, D'Angelo R, Sidoti A. Stargardt Phenotype Associated With Two ELOVL4 Promoter Variants and ELOVL4 Downregulation: New Possible Perspective to Etiopathogenesis? *Invest Ophthalmol Vis Sci.* 2018; 59(2):843-857.

miRNA expression profile of retinal pigment epithelial (RPE) cells under oxidative stress condition

[2018]

<https://febs.onlinelibrary.wiley.com/doi/full/10.1002/2211-5463.12360>

Donato L, Bramanti P, Scimone C, Rianldi C, D'Angelo R, Sidoti A. miRNA expression profile of retinal pigment epithelial (RPE) cells under oxidative stress conditions. *FEBS Open Bio.* 2018; 8(2):219-233.

Two Novel KRIT1 and CCM2 Mutations in Patients Affected by Cerebral Cavernous Malformations

[2018]

<https://www.frontiersin.org/articles/10.3389/fneur.2018.00953/full>

Scimone C, Donato L, Katsarou Z, Bostantjopoulou S, D'Angelo R, Sidoti A. Two Novel KRIT1 and CCM2 Mutations in Patients Affected by Cerebral Cavernous Malformations: New Information on CCM2 Penetrance. *Front Neurol.* 2018; 9:953.

Relevance of CCM gene polymorphisms for clinical management of sporadic cerebral cavernous

[2017]

<https://www.sciencedirect.com/science/article/pii/S0022510X17304185>

Rinaldi C, Bramanti P, Scimone C, Donato L, Alafaci C, D'Angelo R, Sidoti A. Relevance of CCM gene polymorphisms for clinical management of sporadic cerebral cavernous malformations. *J Neurol Sci.* 2017; 380:31-37.

Possible protective role of the ABCA4 gene c.1268A>G missense variant in Stargardt disease

[2017]

<https://www.spandidos-publications.com/10.3892/ijmm.2017.2917>

D'Angelo R, Donato L, Venza I, Scimone C, Aragona P, Sidoti A. Possible protective role of the ABCA4 gene c. 1268A>G missense variant in Stargardt disease and syndromic retinitis pigmentosa in a Sicilian family: Preliminary data. *Int J Mol Med.* 2017; 39(4):1011-1020.

A novel RLBP1 gene geographical area-related mutation present in a young patient with retinitis

[2017]

<https://humgenomics.biomedcentral.com/articles/10.1186/s40246-017-0114-6>

Scimone C, Donato L, Esposito T, Rinaldi C, D'Angelo R, Sidoti A. A novel RLBP1 gene geographical area-related mutation present in a young patient with retinitis punctata albescens. *Hum Genomics.* 2017; 11(1):18.

Update on novel CCM genes mutations in patients with Cerebral Cavernous Malformations

[2016]

<https://link.springer.com/article/10.1007/s12031-016-0863-z>

Scimone C, Bramanti P, Alafaci C, Granata F, Piva F, Rinaldi C, Donato L, Greco F, Sidoti A, D'Angelo R. Update on novel CCM genes mutations in patients with Cerebral Cavernous Malformations. *J Mol Neurosci*. 2016; 61(2): 189-198.

Possible related functions of the non-homologous co-regulated gene pair PDCD10 and SERPINI1

[2017]

http://www.embj.org/wp-content/uploads/2017/04/k2_attachments_Scimone_09.pdf

Scimone C. Possible related functions of the non-homologous co-regulated gene pair PDCD10 and SERPINI1. *Euro Mediterranean Biomedical Journal*. 2017; 12(9):41-46.

CCM3/SERPINI1 bidirectional promoter variants in patients with cerebral cavernous malformations

[2016]

<https://bmcmedgenet.biomedcentral.com/articles/10.1186/s12881-016-0332-0>

Scimone C, Ruggeri A, Bramanti P, Donato L, Alafaci C, Crisafulli C, Mucciardi M, Rinaldi C, Sidoti A, D'Angelo R. CCM3/SERPINI1 bidirectional promoter variants in patients with cerebral cavernous malformations: a molecular and functional study. *BMC Med Genet*. 2016; 17(1):74.

First case of Currarino Syndrome and Trimethylaminuria: two rare diseases for a complex clinical

[2015]

<https://onlinelibrary.wiley.com/doi/full/10.1111/1751-2980.12373>

Scimone C, Donato L, Rinaldi C, Sidoti A, D'Angelo R. First case of Currarino Syndrome and Trimethylaminuria: two rare diseases for a complex clinical presentation. *J Dig Dis*. 2016; 17(9):628-632.

Detection of Novel Mutation in Ccm3 Causes Familial Cerebral Cavernous Malformations

[2015]

<https://europepmc.org/article/med/26115622>

Scimone C, Bramanti P, Ruggeri A, Katsarou Z, Donato L, Sidoti A, D'Angelo R. Detection of Novel Mutation in Ccm3 Causes Familial Cerebral Cavernous Malformations. *J Mol Neurosci*. 2015; 57(3):400-403.

Next Generation Semiconductor Based Sequencing of the Donkey (Equus asinus) Genome Provided

[2015]

<https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0131925>

Bertolini F, Scimone C, Geraci C, Schiavo G, Utzeri VJ, Chiofalo V, Fontanesi L. Next Generation Semiconductor Based Sequencing of the Donkey (Equus asinus) Genome Provided Comparative Sequence Data against the Horse Genome and a Few Millions of Single Nucleotide Polymorphisms. *PLoS One*. 2015; 10(7):e0131925.

Glyoxalase I A111E, Paraoxonase 1 Q192R and L55M polymorphisms in Italian patients with sporadic

[2015]

<https://europepmc.org/article/med/26122242>

Rinaldi C, Bramanti P, Famà A, Scimone C, Donato L, Antognelli C, Alafaci C, Tomasello F, D'Angelo R, Sidoti A. Glyoxalase I A111E, Paraoxonase 1 Q192R and L55M polymorphisms in Italian patients with sporadic Cerebral Cavernous Malformations: a pilot study. *J Biol Regul Homeost Agents*. 2015; Apr-Jun;29(2):493-500.

PON I and GLO I Gene Polymorphisms and Their Association with Breast Cancer: A Case-Control Study

[2014]

<https://www.hilarispublisher.com/abstract/pon-i-and-glo-i-gene-polymorphisms-and-their-association-with-breast-cancer-a-casecontrol-study-in-a-population-from-sou-34691.html>

Rinaldi C, D' Angelo R, Ruggeri A, Calabrò M, Scimone C, Sidoti A. PON I and GLO I Gene Polymorphisms and Their Association with Breast Cancer: A Case-Control Study in a Population from Southern Italy. *Mol Biomark Diagn*. 2014; 5:170.

Fish odor syndrome (trimethylaminuria) supporting the possible FMO3 down expression in childhood

[2014]

<https://jmedicalcasereports.biomedcentral.com/articles/10.1186/1752-1947-8-328>

D'Angelo R, Scimone C, Esposito T, Bruschetta D, Rinaldi C, Ruggeri A, Sidoti A. Fish odor syndrome (trimethylaminuria) supporting the possible FMO3 down expression in childhood: a case report. *J Med Case Rep*. 2014; 8:328.

Sporadic cerebral cavernous malformations: report of further mutations of CCM genes in 40 Italian

[2013]

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CONFERENCES AND SEMINARS

Conference on Genomics and Molecular Biology (GMB-2021)

[Virtual event, 28/09/2021 – 29/09/2021]

Oral communication: [Scimone C](#), Donato L, Alibrandi S, Alafaci C, D'Angelo R, Antonina Sidoti. Editing modifications in Cerebral Cavernous Malformation-derived endothelial cells. *Conference on Genomics and Molecular Biology*. **Virtual event, 28-29 settembre 2021**

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Oral communication: [Scimone C](#), Donato L, Alibrandi S, Caragliano AA, Mormina E, Vinci S, D'Angelo R, Sidoti A. Molecular signaling in sporadic brain arteriovenous malformation. *Virtual event*. **24-27 settembre 2021**

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[Houston, TX, 12/07/2021 – 14/07/2021]

Oral communication: [Scimone C](#), Donato L, Alibrandi S, Caragliano AA, Mormina E, Vinci S, D'Angelo R, Sidoti A. Involvement of imprinted genes in molecular mechanism resulting in pediatric brain arteriovenous malformations. *CEB-2021*. **Houston, TX, USA, 12-14 luglio 2021**

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Organizing Committee Member

International Conference and Exhibition on Genome Science 2019

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Poster presentation: [Scimone C](#), Donato L, D'Angelo R, Sidoti A. Pedigree analysis of a family affected by hereditary cerebral cavernous malformations novel candidate genes detected by whole genome sequencing. *International Conference and Exhibition on Genome Science 2019*, **Houston, USA, September 27-28, 2019**

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ABSTRACT PRESENTATI A CONGRESSI NAZIONALI ED INTERNAZIONALI

Co-autore di oltre 60 abstract presentati a Congressi nazionali ed internazionali

[2010 – Current]

Autorizzo il trattamento dei miei dati personali presenti nel CV ai sensi dell'art. 13 d. lgs. 30 giugno 2003 n. 196 - "Codice in materia di protezione dei dati personali" e dell'art. 13 GDPR 679/16 - "Regolamento europeo sulla protezione dei dati personali".

Messina, 17/03/2022