



### Academic Personnel Short Profile / Short CV

<b>Institution:</b>	The Cyprus Institute of Neurology and Genetics (CING)
<b>Surname:</b>	Stephanou
<b>Name:</b>	Coralea
<b>Rank/Position:</b>	Associate Scientist
<b>Program of Study:</b>	MSc in Molecular Medicine / MSc in Medical Genetics / MSc in Neuroscience / MSc in Biomedical Research / MSc in Biotechnology / PhD in Molecular Medicine / PhD in Medical Genetics / PhD in Neuroscience
<b>Scientific Domain: *</b>	Molecular Biology/Genetics, Genetic modifiers, Clinical variants, Data Curation, Haemoglobinopathies, Rare anaemias

*\*Field of Specialization*

### Academic qualifications / (list by highest qualification)

Qualification	Year	Awarding Institution	Department	Thesis title
PhD in Genetics and Molecular Medicine	2015	King's College London (KCL), London, U.K	Department of Medical and Molecular Genetics	Advancing lentiviral gene-therapy vectors for $\beta$ -thalassaemia
MSc in Genetics of Human Disease	2010	University College London, London, U.K.	Centre for Cardiovascular Genetics, Department of Medicine	Replication study of genetic loci determining telomere length
BSc/ARCS in Biology	2009	Imperial College of Science, Technology and Medicine, London, U.K	Department of Life Sciences	Characterization of eGFP expression in <i>Magnaporthe oryzae</i> under the control of <i>Blumeria graminis</i> f.sp. <i>hordei</i> H4 promoter



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ΦΟΡΕΑΣ ΔΙΑΣΦΑΛΙΣΗΣ ΚΑΙ ΠΙΣΤΟΠΟΙΗΣΗΣ ΤΗΣ ΠΟΙΟΤΗΤΑΣ ΤΗΣ ΑΝΩΤΕΡΗΣ ΕΚΠΑΙΔΕΥΣΗΣ  
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### Employment history in Academic Institutions/Research Centers – List by the three (3) most recent

Period of employment		Employer	Location	Position
From	To			
March 2021	Present	Molecular Genetics Thalassaemia Department, The Cyprus Institute of Neurology and Genetics	Nicosia, Cyprus	Associate Scientist
Sept. 2021	Present	The Cyprus Institute of Neurology and Genetics	Nicosia, Cyprus	Associate Faculty
March 2019	Febr. 2021	Molecular Genetics Thalassaemia Department, The Cyprus Institute of Neurology and Genetics	Nicosia, Cyprus	Laboratory Scientific Officer
Sept. 2017	Febr. 2019	Agrobiotechnology Department, Agricultural Research Institute Project title: OLIVE-MIRACLE – Modeling solutions for improved and Resilient management strategies for olive tree against future climate change	Nicosia, Cyprus	Post-doctoral Fellow
Dec. 2016	Dec. 2018	Molecular Genetics Thalassaemia Department, The Cyprus Institute of Neurology and Genetics	Nicosia, Cyprus	Part-time Laboratory Scientific Officer
March 2016	Oct. 2016	Molecular Genetics Thalassaemia Department, The Cyprus Institute of Neurology and Genetics Project title: ThalaMoSS - Thalassaemia Modular Stratification System for Personalized Therapy of $\beta$ -Thalassemia	Nicosia, Cyprus	Post-doctoral Fellow
May 6, 2016	June 17, 2016	Weill Cornell Medical College	N.Y., U.S.A.	Visiting Researcher



Period of employment		Employer	Location	Position
From	To			

**Key refereed journal papers, monographs, books, conference publications etc.  
List the five (5) more recent and other five (5) selected –(max total 10)**

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
1	2023	IthaPhen: An Interactive Database of Genotype-Phenotype Data for Hemoglobinopathies	Xenophontos M, Minaidou A, <b>Stephanou C</b> , Tamana S, Kleanthous M, Kountouris P.	Journal: Hemasphere Publisher: Wolters Kluwer Health	7	e922
2	2022	A Novel Tool for the Analysis and Detection of Copy Number Variants Associated with Haemoglobinopathies.	Minaidou A, Tamana S, <b>Stephanou C</b> , Xenophontos M, Hartevelde CL, Bento C, Kleanthous M <sup>§</sup> , Kountouris P <sup>§</sup> .	Journal: International Journal of Molecular Sciences Publisher: MDPI	23	15920
3	2022	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies	Tamana S <sup>‡</sup> , Xenophontos M <sup>‡</sup> , Minaidou A, <b>Stephanou C</b> , Hartevelde CL, Bento C, Traeger-Synodinos J, Fylaktou I, Yasin NM, Abdul Hamid FS, Esa E, Halim-Fikri H, Zilfalil BA, Kakouri AC; ClinGen Hemoglobinopathy Variant Curation Expert Panel; Kleanthous M, Kountouris P.	Journal: Elife Publisher: eLife Sciences Publications Ltd	11	e7971 3

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
4	2022	Optimized Droplet Digital PCR Assay on Cell-Free DNA Samples for Non-Invasive Prenatal Diagnosis: Application to Beta-Thalassemia	Constantinou CG, Karitzi E, Byrou S, <b>Stephanou C</b> , Michailidou K, Makariou C, Hadjilambi G, Christofides A, Kleanthous M, Papasavva T.	Journal: Clinical Chemistry Publisher: Oxford University Press	68	1053-1063
5	2022	Global Globin Network Consensus Paper: Classification and Stratified Roadmaps for Improved Thalassaemia Care and Prevention in 32 Countries	Halim-Fikri BH, Lederer CW, Baig AA, Mat-Ghani SNA, Syed-Hassan SR, Yusof W, Abdul Rashid D, Azman NF, Fucharoen S, Panigoro R, Silao CLT, Viprakasit V, Jalil N, Mohd Yasin N, Bahar R, Selvaratnam V, Mohamad N, Nik Hassan NN, Esa E, Krause A, Robinson H, Hasler J, <b>Stephanou C</b> , Raja-Sabudin RZ, Elion J, El-Kamah G, Coviello D, Yusoff N, Abdul Latiff Z, Arnold C, Burn J, Kountouris P, Kleanthous M, Ramesar R, Zilfalil BA, On Behalf Of The Global Globin Network Ggn.	Journal: Journal of Personalised Medicine Publisher: MDPI	12	552
6	2021	The International Hemoglobinopathy Research Network (INHERENT): an international initiative to	Kountouris P, <b>Stephanou C</b> , Archer N, Bonifazi F, Giannuzzi V, Kuo KHM, Maggio A, Makani J, Mañú-Pereira MDM,	Journal: American Journal of Hematology Publisher: Wiley, Wiley Liss Inc	96	E416-E420

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
		study the role of genetic modifiers in hemoglobinopathies	Michailidou K, Nkya S, Nnodu O, Trompeter S, Tshilolo L, Wonkam A, Zilfalil BA, Inusa BPD, and Kleanthous M, on behalf of the International Hemoglobinopathy Research Network (INHERENT).			
7	2021	Adapting the ACMG/AMP variant classification framework: a perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel	Kountouris P <sup>‡</sup> , <b>Stephanou C<sup>‡</sup></b> , Lederer CW, Traeger-Synodinos J, Bento C, Hartevelde CL, Fylaktou E, Koopmann TT, Halim-Fikri H, Michailidou K, Nfonsam LE, Waye JS, Zilfalil BA, and Kleanthous M, on behalf of the ClinGen Hemoglobinopathy Variant Curation Expert Panel.	Journal: Human Mutation Publisher: Wiley	43	1089-1096
8	2021	Land use in urban areas impacts the composition of soil bacterial communities involved in nitrogen cycling. A case study from Lefkosia (Nicosia) Cyprus	<b>Stephanou C</b> , Omirou M, Philippot L, Zissimos AM, Christoforou IC, Trajanoski S, Oulas A, Ioannides IM.	Journal: Scientific Reports Publisher: Nature Portfolio	11	8198
9	2019	Genetic modifiers at the crossroads of personalized	<b>Stephanou C</b> , Tamana S, Minaidou A,	Journal: Journal of Clinical Medicine Publisher: MDPI	8	1927

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
		medicine for haemoglobinopathies	Papasavva P, Kleanthous M, Kountouris P.			
10	2017	Suitability of Small Diagnostic Peripheral-blood Samples for Cell-therapy Studies.	<b>Stephanou C</b> , Papasavva P, Zachariou M, Patsali P, Epitropou M, Christou S, Antoniou M, Lederer CW, Kleanthous M.	Journal: Cytotherapy Publisher: Elsevier	19	311-326

**Exhibitions (where applicable). List the five (5) more recent and other five (5) selected. (max total 10)**

Ref. No.	Date	Topic	International / Local	Location*	Role in Exhibition
1	24 October 2023	ARISE General Assembly and Steering Committee meeting	International	London, U.K.	Oral Presentation
2	25-28 October 2023	The 18th Annual Sickle Cell & Thalassemia Conference (ASCAT)	International	London, U.K.	Poster Presentation
3	27-28 January 2022	ASCAT 2022: 2nd Virtual Sickle Cell & Thalassemia Conference in collaboration with EHA & BSH	International	Virtual	Poster Presentation
4	24-25 June 2021	ClinGen Virtual Retreat 2021	International	Virtual	Poster Presentation
5	12-14 May 2021	5th Curating the Clinical Genome conference	International	Virtual	Poster Presentation
6	4-5 December 2020	8th International Conference of the Cyprus Society of Human Genetics	Local	Virtual	Poster Presentation
7	6-9 June 2020	European Human Genetics Conference	International	Virtual	Poster Presentation
8	7-9 November 2019	7th International Multithematic Bio-Medical Congress	Local	Nicosia, CY	Poster Presentation



Ref. No.	Date	Topic	International / Local	Location*	Role in Exhibition
9	28-30 October 2019	Global Globin 2020 (GG2020) Challenge Conference	International	UNESCO, Paris, France	Invited Oral & Poster Presentation
10	16-18 November 2012	3rd International Conference of the Cyprus Society of Human Genetics	Local	Nicosia, CY	Poster Presentation

\* Specify venue, geographic location etc

**Research Projects. List the five (5) more recent and other five (5) selected  
(max total 10)**

Ref. No.	Date	Title	Funded by	Project Role*
1	Sept. 2027	Haemoglobinopathies in European Liaison of Medicine and Science (HELIOS) <a href="https://www.cost.eu/actions/CA22119/">https://www.cost.eu/actions/CA22119/</a>	EU Framework Programmes for research and innovation	Working Group co-Leader Core Group member Candidate MC member
2		Study of the role of genetic modifiers in $\beta$ -thalassemia patients in Cyprus (EEBK/ΕΠ/2021/57)	CING	Co-investigator
3		ITHANET Portal <a href="https://www.ithanet.eu/home/ithanet-project">https://www.ithanet.eu/home/ithanet-project</a>	FP6/CING	Member of Project Team Data curator
4		International Hemoglobinopathy Research Network <a href="https://www.inherentnetwork.org/">https://www.inherentnetwork.org/</a>	CING/industry	Member of Project Team Steering Committee Member Working Group Leader
5		European Reference Network on Rare Haematological Diseases (ERN-EuroBloodNet) <a href="https://eurobloodnet.eu/ern-eurobloodnet/">https://eurobloodnet.eu/ern-eurobloodnet/</a>		Member of Project Team
6		ClinGen Hemoglobinopathy Variant Curation Expert Panel (VCEP) <a href="https://clinicalgenome.org/affiliation/50052/">https://clinicalgenome.org/affiliation/50052/</a>	CING	Panel Member and Biocurator Trainer
7	Oct. 2024	African Research and Innovative Initiative for Sickle cell Education (ARISE) <a href="https://www.ariseinitiative.org/">https://www.ariseinitiative.org/</a>	EU Horizon 2020-MSCA-RISE	Member of Project Team Staff Exchange Coordinator at the CING
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\*Project Role: i.e. Scientific/Project Coordinator, Research Team Member, Researcher, Assistant Researcher, other

**Academic Consulting Services and/or Participation in Councils / Boards/ Editorial Committees.  
List the five (5) more recent (Optional Entry)**

Ref. No.	Period	Organization	Title of Position or Service	Key Activities
1	Sept. 2021 - Present	The Cyprus Institute of Neurology and Genetics	Associate Faculty	Lectures and workshops Postgraduate course "MM101 – Molecular Basis of Monogenic Diseases"  MSc thesis examination committee member; PhD project co-supervisor
2	June 2019 - Present	Staff exchange programme ARISE ( <a href="https://www.ariseinitiative.org/">https://www.ariseinitiative.org/</a> )	Staff Exchange Coordinator	Coordinate the activities for staff exchanges at the CING, including the preparation/review of study proposals, laboratory supervision, presentations at project meetings, and sorting out visa and other contractual issues.
3	Present	Biomedicines Special Issue "Hemoglobin Molecule: A Structure-Function Interplay" (MDPI ISSN 2227-9059)	Co-Guest Editor	
4	4 March 2023	12 <sup>th</sup> Pancyprrian Thalassaemia Conference	Conference organizing committee member	Participate in the initial planning stages to define the conference's goals, objectives, and target audience; Contribute to the creation of the conference program; Schedule sessions and plan breaks;



Ref. No.	Period	Organization	Title of Position or Service	Key Activities
				Help establish budget strategy and reach out to potential sponsor organizations with a personalized e-mail; Keep track of financial transactions; Coordinate with the venue staff for logistical arrangements, such as room setups, audiovisual equipment, catering; Be present at the conference to address any issues that may arise; Oversee the smooth execution of the event; Act as the Session Moderator; Maintain records of all conference-related activities, including invoices and correspondence; Prepare and send via e-mail conference attendance certificates; Prepare and publish a lay article on the activities and key outcomes of the conference
5	2021/2022	ClinGen Hemoglobinopathy Variant Curation Expert Panel (VCEP)	Biocurator Trainer; coordinate activities of biocurators for the timely completion of the pilot study	Define the specific goals and objectives of the pilot stud; Create a detailed timeline; Establish clear procedures and guidelines for data curation; Schedule regular online meetings to monitor progress and address any challenges or questions; Foster open communication among the biocurators, enabling them to collaborate and share insights;



Ref. No.	Period	Organization	Title of Position or Service	Key Activities
				Maintain detailed documentation of the study's progress, including data curation processes and any changes made; Conduct a final review of the pilot study to ensure that all tasks have been completed as planned; Prepare a summary report
	2021 - Present		Peer-reviewer	Peer-review activity for Genetic Testing and Molecular Biomarkers, Journal of Clinical Pathology, Hemoglobin

**Awards / International Recognition (where applicable). List the five (5) more recent and other five (5) selected. (max total 10) (Optional Entry)**

Ref. No.	Date	Title	Awarded by:
1	October 2019	Best Poster Award (1st Prize), "ITHANET: An information and database community portal for haemoglobinopathies"	Global Globin 2020 (GG2020) Challenge Conference, UNESCO, Paris, France
2	2010	UGI Prize–Student of the Year 2009–2010	University College London, U.K.
3	June 2005	GCE O'levels award for academic excellence	The Grammar School Parents and Guardians Association
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ΦΟΡΕΑΣ ΔΙΑΣΦΑΛΙΣΗΣ ΚΑΙ ΠΙΣΤΟΠΟΙΗΣΗΣ ΤΗΣ ΠΟΙΟΤΗΤΑΣ ΤΗΣ ΑΝΩΤΕΡΗΣ ΕΚΠΑΙΔΕΥΣΗΣ  
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Ref. No.	Date	Title	Awarded by:
10			

**Other Achievements. List the five (5) more recent and other five (5) selected.  
 (max total 10) (Optional Entry)**

Ref. No.	Date	Title	Key Activities:
1	2019	"NIPD of beta-Globinopathies using Targeted Next Generation Sequencing Haplotype Analysis", RPF POST-DOC/0916/0238, funded (score 14.10/15.00) with €160,000	Awarded a post-doctoral fellowship; not pursued
2	2010	"The Community Assessment of Community Annotation with Ontologies (CACAO) Competition" <a href="https://gowiki.tamu.edu/wiki/index.php/Category:CACAO_Fall_2010">https://gowiki.tamu.edu/wiki/index.php/Category:CACAO_Fall_2010</a>	Multi-institution competition for teams of undergraduate students around the world to improve GO annotation of genes; manual annotations. Participated with the UCL Institute Of Cardiovascular Science (U.K.)
3		HAPLONID (Non-Invasive Prenatal Diagnosis of beta-Globinopathies using Targeted Next Generation Sequencing Haplotype Analysis), RPF EXCELLENCE/1216/0484, funded (score 14.25/15.00) with €250,000.	Co-investigator (Proposal Write-up)
4		THALAMOSS (THALAssaemia MOdular Stratification System for personalized therapy of beta-thalassemia), EC FP7 grant agreement no. 306201, funded with €5,02 Mio total (€581,725 Cyprus)	Co-investigator (Experimentally)
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