



### Academic Personnel Short Profile / Short CV

<b>Institution:</b>	The Cyprus Institute of Neurology and Genetics (CING)
<b>Surname:</b>	Papasavva
<b>Name:</b>	Thessalia
<b>Rank/Position:</b>	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>	Monogenic Disorders/Genetic Haemoglobinopathy

*\*Field of Specialization*

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

Qualification	Year	Awarding Institution	Department	Thesis title
PhD	2012	National and Kapodistrian University of Athens	Medical School, Greece	Development of a Non-Invasive Prenatal Diagnostic Method for $\beta$ -thalassaemia using SNPs.
MSc	1996	University College London (UCL), UK.	Applied Molecular Biology and Biotechnology	
BSc	1995	University of Arizona, USA	Microbiology	



ΦΟΡΕΑΣ ΔΙΑΣΦΑΛΙΣΗΣ ΚΑΙ ΠΙΣΤΟΠΟΙΗΣΗΣ ΤΗΣ ΠΟΙΟΤΗΤΑΣ ΤΗΣ ΑΝΩΤΕΡΗΣ ΕΚΠΑΙΔΕΥΣΗΣ  
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Qualification	Year	Awarding Institution	Department	Thesis title

**Employment history in Academic Institutions/Research Centers – List by the three (3) most recent**

Period of employment		Employer	Location	Position
From	To			
2004	Present	The Cyprus Inst of Neurology and Genetics	Molecular Genetics Thalassaemia Dep	Scientist
2001	2003	The Cyprus Inst of Neurology and Genetics	Electron Microscopy and Molecular Pathology Department	Laboratory Scientific Officer
Jan 2001	July 2001	Kyperouna Hospital Lab	Clinical Lab	Laboratory Scientific Officer
1996	2001	The Cyprus Inst of Neurology and Genetics	Molecular Virology Department	Laboratory Scientific Officer

**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	2024	Unravelling the Complexity of the +33 C>G [HBB:c.-18C>G] Variant in Beta Thalassaemia	Citation: Stephanou, C.; Petrou, M.; Kountouris, P.; Makariou, C.; Christou, S.; Hadjigavriel, M.; Kleanthous, M.; Papasavva, T.	Biomedicines	12	1-13

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
2	2022	Optimized Droplet Digital PCR Assay on Cell-Free DNA Samples for Non-Invasive Prenatal Diagnosis: Application to Beta-Thalassemia	<ul style="list-style-type: none"> <li>Constantina G Constantinou, Eleni Karitzi, Stefania Byrou, Coralea Stephanou, Kyriaki Michailidou,</li> </ul> Christiana Makariou, Georgia Hadjilambi, Agathoklis Christofides, Marina Kleanthous*, and Thessalia Papasavva*	Clinical Chemistry	27;68(8):	1053-1063
3	2022	Direct Chromosomal Phasing: An Easy and Fast Approach for Broadening Prenatal Diagnostic Applicability	Stefania Byrou, George Christopoulos, Agathoklis Christofides, Christiana Makariou, Christiana Ioannou, Marina Kleanthous*, Thessalia Papasavva	Thalassemia Reports	12,3	55-72
4	2022	THE CLINGEN HEMOGLOBINOPATHY VARIANT CURATION EXPERT PANEL	C Coralea Stephanou, P Petros Kountouris, C Carsten W Lederer, C Celeste Bento, C Cornelis L Hartveld, J Jan Traeger-Synodinos, J John S Waye, Z Zhiyu Peng, I Irene Fylaktou, H Hashim Halim-Fikri, T Tamara T Koopmann, L Landry	Hemasphere	6	S1

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
			Nfonsam, J Jun Sun, F Franck Nzengu-Lukusa, M Michael Angastiniotis, C Catherine Badens, B Bertha Ibarra Cortes, J Johan T den Dunnen, J Jacques Elion, S Suthat Fucharoen, K Kyriaki Michailidou, T Thessalia Papasavva, A Antonio Piga, R Raj Ramesar, S Swee Lay Thein, L Léon Tshilolo, Z Zilfalil Bin Alwi, M Marina Kleanthous			
5	2018	Fast Temperature-Gradient COLD PCR for the enrichment of the paternally inherited SNPs in cell free fetal DNA; an application to non-invasive prenatal diagnosis of $\beta$ - thalassaemia.	Byrou S, Makrigiorgos GM, Christofides A, Kallikas I, Papasavva T*, Kleanthous M	PlosOne	25	13
6	2016	Prevalence of RhD status and clinical application of non-invasive prenatal determination of fetal RHD	Thessalia Papasavva , Pete Martin , Tobias J Legler , Marios Liasides , George Anastasiou , Agathoklis Christofides ,	BMC Res Notes	1	198

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
		in maternal plasma: a 5 year experience in Cyprus	Tasos Christodoulou , Sotos Demetriou , Prokopis Kerimis , Charis Kontos 1, George Leontiades , Demetris Papapetrou , Telis Patroclos , Marios Phylaktou , Nikos Zottis 1, Eleni Karitzie , Eleni Pavlou , Petros Kountouris, Barbera Veldhuisen, Ellen van der Schoot, Marina Kleanthous			
7	2016	The molecular spectrum and distribution of haemoglobinopathies in Cyprus: a 20-year retrospective study	Kountouris P, Kousiappa I, Papasavva T, et al	Scientific Reports	6	26371
8	2013	Next generation sequencing of SNPs for non-invasive prenatal diagnosis: challenges and feasibility as illustrated by an application to $\beta$ - thalassaemia	Thessalia Papasavva, Wilfred F J van IJcken, Christel E M Kockx, Mirjam C G N van den Hout, Petros Kountouris, Loukas Kythreotis, Eleni Kalogirou, Frank G Grosveld, and Marina Kleanthous	<i>Eur J Hum Genet</i>	21	1403



Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
9	2013	A minimal set of SNPs for the non-invasive prenatal diagnosis of $\beta$ -thalassaemia	Thessalia E Papasavva, Carsten W Lederer, Jan Traeger-Synodinos, Ariadne Mavrou, Emmanuel Kanavakis, Christiana Ioannou, Christiana Makariou, Marina Kleanthous	Annals of Human Genetics	77	115-124
10	2010	ThalassoChip, an array mutation and single nucleotide polymorphism detection tool for the diagnosis of $\beta$ -thalassaemia	Christos Shammias, Thessalia Papasavva, Xenia Felekis, Christos Christophorou, Hanno Roomere, Jan Traeger Synodinos, Emmanuel Kanavakis, Mohammed El-Khateeb, Hanan Hamamy, Tamara Mahmoud, Mohammad Shboul, Amal El Beshlawy, Dvora Filon, Ibtesam R Hussein, Renzo Galanello, Giovanni Romeo, Marina Kleanthous	<i>Clinical Chemistry and Laboratory Medicine</i>	48	1713

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
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*\*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	01/09/2019- 31/01/2023	<i>Non-Invasive Prenatal Diagnosis of <math>\beta</math>-Globinopathies using Targeted Next Generation Sequencing Haplotype Analysis</i>	RPF RESTART 2016 – 2020	Project Coordinator
2	2021-2022	Shielding the Next Generation with Preconception Genetic Screening,	CING	Project Coordinator
3	2016-2019	Protecting the Health of the Next Generation with Preconception Genetic Screening (PCGS)	RCB	Project Coordinator
4	2004-2008	SAFE: Special Non-Invasive Advances in Foetal and Neonatal Evaluation Network,	FP6-LSHB-CT- 2004-503243	Researcher, PhD student
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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	2017-present	EMQN/GenQA	Member of the EQA-NIPT Assessing committee	Assessor of EQA schemes
2	2021-2023	Cyprus Society of Human Genetics	Elected Board Member, Secretary	
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**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:
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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	January 2023	A patent application for our work in Non-Invasive Prenatal Diagnosis for $\beta$ -haemoglobinopathies is filed: Filing date:16.01.2023, Ref No: CYPBB/P80598GB, Patent Application No:2300597.8	
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