



### Academic Personnel Short Profile / Short CV

<b>Institution:</b>	The Cyprus Institute of Neurology & Genetics (CING)
<b>Surname:</b>	Drousiotou
<b>Name:</b>	Anthi
<b>Rank/Position:</b>	Professor, Emeritus Senior Scientist and Founder, Biochemical Genetics Department, The Cyprus Institute of Neurology & Genetics
<b>Program of Study:</b>	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>	Biochemical Genetics

### Academic qualifications (list by highest qualification)

Qualification	Year	Awarding Institution	Department	Thesis title
PhD	1983	University of London	Department of Biochemistry, Guy's Hospital Medical School	Is the erythrocyte affected in Duchenne Muscular Dystrophy?
BSc	1979	Imperial College, University of London	Biochemistry	



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

Period of employment		Employer	Location	Position
From	To			
2003	2022	The Cyprus Institute of Neurology & Genetics	The Cyprus Institute of Neurology & Genetics	Head of the Department of Biochemical Genetics
1989	2003	The Cyprus Government	Thalassaemia Centre, Nicosia	Senior Laboratory Officer
1984	1989	The Cyprus Government	Limassol General Hospital	Laboratory 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. Number	Year	Title	Other authors	Journal and Publisher/ Conference	Vol.	Pages
1	2023	“GAA variants associated with reduced enzymatic activity but lack of Pompe-related symptoms, incidentally identified by exome sequencing”	A Malekkou, A Theodosiou, A Alexandrou, I Papaevripidou, C Sismani, E H Jacobs, G J G Ruijter, V Anastasiadou, S Ourani, E Athanasiou, <b>A Drousiotou</b> , O Grafakou, P P Petrou	Mol Genet Metab Rep	36	doi: <a href="https://doi.org/10.1016/j.yimgmr.2023.100997">10.1016/j.yimgmr.2023.100997</a>
2	2022	“Newly identified disorder of copper metabolism caused by variants in CTR1, a high-affinity copper transporter”	S Batzios, G Tal, A T DiStasio, Y Peng, C Charalambous, P Nicolaides, E-J Kamsteeg, S H Korman, H	Hum Mol Genet	16;31 (24)	4121-4130



			Mandel, P J Steinbach, L Yi, S R Fair, M E Hester, <b>A Drousiotou</b> S G Kaler	.		
3	2022	“Stbd1-deficient mice display insulin resistance associated with enhanced hepatic ER-mitochondria contact”	S Kyriakoudi A Theodoulou, L Potamiti, F Schumacher, M Zachariou, R Papacharalambous, B Kleuser, M I Panayiotidis, <b>A Drousiotou</b> , P P Petrou	Biochimie	200	172-183
4	2021	“When the Balance Tips: Dysregulation of Mitochondrial Dynamics as a Culprit in Disease”	Kyriakoudi S, <b>Drousiotou A</b> , Petrou PP.	Int. J. Mol. Sci.	22(9)	4617
5	2020	“Stbd1 promotes glycogen clustering during endoplasmic reticulum stress and supports survival of mouse myoblasts”	A A Lytridou, A Demetriadou, M Christou, L Potamiti, N P Mastrogiannopoulos, K Kyriacou, L A Phylactou, <b>A Drousiotou</b> , Petros P Petrou	J Cell Sci	133 (20)	doi:10.1242/jcs.244855
6	2020	“Acid Ceramidase Depletion Impairs Neuronal Survival and Induces Morphological Defects in Neurites Associated with Altered Gene Transcription and Sphingolipid content”	K Kyriakou, CW Lederer, M Kleanthous, <b>A Drousiotou</b> and A Malekkou.	Int. J. Mol. Sci.	21(5)	doi:10.3390/ijms21051607



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7	2019	"A novel mutation deep within intron 7 of the <i>GBA</i> gene causes Gaucher disease"	Malekku A, Sevastou I, Mavrikiou G, Georgiou T, Vilageliu L, Moraitou M, Michelakakis H, Prokopiou C, <b>Drousiotou A.</b>	Mol Genet Genomic Med.	8(3)	<a href="http://doi.org/10.1002/mgg3.1090">http://doi.org/10.1002/mgg3.1090</a>
8	2019	"Classic galactosaemia in the Greek Cypriot population: An epidemiological and molecular study"	R. Papachristoforou, P. Petrou, H. Sawyer, M. Williams, <b>A. Drousiotou.</b>	Ann Hum Genet	83	291-298
9	2018	"Biochemical Characterization of the <i>GBA2</i> c.1780G>C Missense Mutation in Lymphoblastoid Cells from Patients with Spastic Ataxia"	Malekku A, Samarani M, <b>Drousiotou A</b> , Votsi C, Sonnino S, Pantzaris M, Chiricozzi E, Zamba-Papanicolaou E, Aureli M, Loberto N, Christodoulou K.	Int J Mol Sci.	19(10)	doi:10.3390/ijms19103099.
10	2017	"Mouse <i>Stbd1</i> is N-myristoylated and affects ER-mitochondria association and mitochondrial morphology"	A Demetriadou, J Morales-Sanfrutos, M Nearchou, O Baba, K Kyriacou, EW Tate, <b>A Drousiotou</b> and PP Petrou.	J Cell Sci.	130 (5)	903-915

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

Ref. Number	Date	Topic	International / Local	Location*	Role in Exhibition
1	-	-	-	-	-

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



<b>(max total 10)</b>				
<b>Ref. Number</b>	<b>Date</b>	<b>Title</b>	<b>Funded by</b>	<b>Project Role*</b>
1	2019-2022	“Linking glycogen metabolism and endoplasmic reticulum stress to obesity and diabetes through the glycogen-binding protein Stbd1”	Cyprus Research Promotion Foundation	Research Team member
2	2015-2018	“An investigation into the function of acid ceramidase in motor neurons and its association with Spinal Muscular Atrophy SMA-PME”	Telethon Cyprus	Research Team member
3	2013-2016	“Insights into the mechanisms of lysosomal glycogen transport and implications for Pompe disease”	Telethon Cyprus	Research Team member
4	2011-2012	“The role of endoplasmic reticulum and oxidative stress in the pathogenesis of galactosaemia”	Cyprus Research Promotion Foundation	Scientific / Project Co-ordinator
5	2011-2014	“Molecular characterization of Cypriot patients with glutaric aciduria type I”	Cyprus Institute of Neurology & Genetics	Scientific / Project Co-ordinator
6	2007-2009	“Epidemiology and molecular basis of galactosaemia in Cyprus”	Telethon Cyprus	Scientific / Project Co-ordinator
7	2010-2012	“Molecular characterization of Cypriot patients with phenylalanine hydroxylase deficiency detected through neonatal screening”	Cyprus Institute of Neurology & Genetics	Scientific / Project Co-ordinator



8	2005-2009	“Molecular characterization of MSUD in Cyprus”	Cyprus Institute of Neurology & Genetics	Scientific / Project Co-ordinator
9	1992-1998	“Neonatal Screening for Duchenne Muscular Dystrophy in Cyprus”	Cyprus Institute of Neurology & Genetics	Scientific / Project Co-ordinator
10	1994-2000	“Epidemiology and molecular characterization of Sandhoff disease in Cyprus”	Cyprus Institute of Neurology & Genetics	Scientific / Project Co-ordinator

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

Ref. Number	Period	Organization	Title of Position or Service	Key Activities
1	2010-present	EuroGentest Project	National Representative for Cyprus in the field of Biochemical Genetics	
2	2012-present	ERNDIM (European Research Network for evaluation and improvement of screening, Diagnosis and treatment of Inherited disorders of Metabolism)	Member of the Board of Trustees	



3	2004-2006 2009-2010	The Cyprus Society of Human Genetics	Founding Member and past Member of the Board of Directors	
4	2010-present	National Committee for Rare Disorders	Member	
5	2011-present	National Committee for Newborn Screening	Member	

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

Ref. Number	Date	Title	Awarded by:
1	1979	Associate of the Royal College of Science	The Royal College of Science, UK

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

Ref. Number	Date	Title	Key Activities:
1	-	-	-



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