



Academic Personnel Short Profile / Short CV

Institution:	The Cyprus Institute of Neurology and Genetics (CING)
Surname:	Sismani
Name:	Carolina
Rank/Position:	Professor
Program of Study:	MSc in Molecular Medicine / MSc in Medical Genetics / MSc in Neuroscience / MSc in Biomedical Research / PhD in Molecular Medicine / PhD in Medical Genetics / PhD in Neuroscience
Scientific Domain: *	Cytogenomics

**Field of Specialization*

Academic qualifications / (list by highest qualification)

Qualification	Year	Awarding Institution	Department	Thesis title
PhD in Human Genetics	2004	Nottingham University	School of Life Sciences	Genetic factors in Mental Retardation
BSc in Biology	1994	Stockholm's University	Biology/Zoology	Effects of different androgens and aromatase inhibitors on secondary sexual characters and steroidogenesis in the testes of male three-spined stickleback."



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			
2012	To date	Cyprus Institute of Neurology and Genetics	Nicosia	Current Professor Programme Coordinator Medical Genetics Course Coordinator MG102
2017	2019	University of Nicosia	Nicosia	Clinical Associate
2010	2010	University of Cyprus	Nicosia	Part time lecturer
1995	1996	Higher Technical Institute	Nicosia	Lecturer

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	Sodium Channel Gene Variants in Fetuses with Abnormal Sonographic Findings: Expanding the Prenatal Phenotypic	Hadjipanteli A, Theodosiou A, Papaevripidou I, Evangelidou P, Alexandrou A, Salameh N, Kallikas I, Kakoullis K,	Genes (Basel), MDPI	15(1)	doi: 10.3390/genes15010119

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
		Spectrum of Sodium Channelopathies..	Frakala S, Oxinou C, Marnerides A, Kousoulidou L, Anastasiadou VC, Sismani C.			
2	2023	CHD2 pathogenic nonsense variant in a three-generation family with variable phenotype and a paracentric inversion 16: Case report	Angelopoulou E, Theodosiou A, Papaevripidou I, Alexandrou A, Liehr T, Gyftodimou Y, Stefanou EG, Sismani C	Helion Journal, Elsevier	9(12)	e2298 7. doi
3	2023	Hereditary multiple exostoses caused by a chromosomal inversion removing part of EXT1 gene	Alexandrou A, Salameh N, Papaevripidou I, Nicolaou N, Myrianthopoulos P, Ketoni A, Kousoulidou L, Anastasiou AM, Evangelidou P, Tanteles GA, Sismani C	Molecular Cytogenetics, Springer	16 (1)	8. doi: 10.11 86/s1 3039- 023- 00638 - 0.PMI D: 37217 936
4	2023	GAA variants associated with reduced enzymatic activity but lack of Pompe-related symptoms, incidentally identified by exome sequencing.	Malekkou A, Theodosiou A, Alexandrou A, Papaevripidou I, Sismani C , Jacobs EH, Ruijter GJG, Anastasiadou V, Ourani S, Athanasiou E, Drousiotou A, Grafakou O, Petrou PP	Molecular Genetics and Metabolism Reports, Elsevier	36:	10099 7. doi: 10.10 16/j.y mgmr. 2023. 10099 7.

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
						PMID: 37600 231
5	2022	Exploring the genetic causality of discordant phenotypes in familial apparently balanced translocation cases using whole exome sequencing	Aristidou C, Theodosiou A, Alexandrou A, Papaevripidou I, Evangelidou E, Kosmaidou-Aravidou Z, Behjati F 3, Christophidou-Anastasiadou V, Tanteles GA and Sismani C	Genes (Basel), MDPI	14(1)	82. doi: 10.3390/genes14010082.
6	2021	Unravelling the genetic causes of multiple malformation syndromes: A whole exome sequencing study of the Cypriot population	Kritioti E, Theodosiou A, Parpaite T, Alexandrou A, Nicolaou N, Papaevripidou I, Séjourné N, Coste B, Christophidou-Anastasiadou V, Tanteles GA, Sismani C	PLoS One, PUBLIC LIBRARY SCIENCE	16(7):	e0253562. doi10.1371/journal.pone.0253562. eCollection 2021. PMID: 34324503
7	2020	Novel Pericentric Inversion inv(9)(p23q22.3) in Unrelated Individuals With	Sismani C, Rapti SM, Iliopoulou P, Spring A, Neroutsou R, Lagou M,	J Hum Genet, Springer	65(9).	783-795

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
		Fertility Problems in the Southeast European Population. J Hum Genet. 2020 May 13.doi: 10.1038/s10038-020-0769-z	Robola M , Tsitsopoulos E, Kousoulidou L , Alexandrou A, Papaevripidou I, Theodosiou A , Syrrou M, Fuchs S, Hempel M , Huhle D , Liehr T, Ziegler M, Duesberg M , Velissariou V			
8	2020	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases	Aristidou C, Theodosiou A, Bak M, Mehrjouy MM, Constantinou E, Alexandrou A, Papaevripidou I, Christophidou-Anastasiadou V, Skordis N, Kitsiou-Tzeli S, Tommerup N, Sismani C	PLoS One, PUBLIC LIBRARY SCIENCE	13(10).	e0205298. doi: 10.1371/journal.pone.0205298
9	2018	De novo unbalanced translocations have a complex history/aetiology	Bonaglia MC, Kurtas NE, Errichiello E, Bertuzzo S, Beri S, Mehrjouy MM, Provenzano A, Vergani D, Pecile V, Novara F, Reho P, Di Giacomo MC, Discepoli G, Giorda R, Aldred MA, Santos-Rebouças CB, Goncalves AP, Abuelo DN, Giglio S, Ricca I, Franchi F, Patsalis P, Sismani C , Morí MA, Nevado J, Tommerup N, Zuffardi O.	Human Genetics, Springer	137(10)	817-829

Ref. No.	Year	Title	Other authors	Journal and Publisher / Conference	Vol.	Pages
10	2017	Accurate breakpoint mapping in apparently balanced translocation families with discordant phenotypes using whole genome mate-pair sequencing.	Aristidou C, Koufaris C, Theodosiou A, Bak M, Mehrjouy MM, Farkhondeh B, Tanteles G, Anastasiadou V, Tommerup N, Sismani C	PLoS One, PUBLIC LIBRARY SCIENCE	12(1)	e0169935. doi: 10.1371/journal.pone.0169935. eCollection 2017. PMID: 28072833

**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	September 2023	International Federation of Fertility Societies, 2023 World Congress,	International	Athens	Presenter
2	June 2023	European Human Genetics Conference	European	Glaskow/Hybrid	Presenter
3	May 2023	EMQN Assessor Meeting	International	Berlin	Laboratory Assessor
4	March 2023	9th International Conference of the Cyprus Society of Human Genetics	Local	Nicosia	Presenter
5	June 2022	European Human Genetics Conference	European	Vienna/Hybrid	Presenter
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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	2013-2016	Establishment of a translational facility at the CING	Norway Government through the Cyprus Planning Bureau	Scientific Co-ordinator
2	2014-2017	Accurate Mapping of breakpoints in balanced translocations	Telethon Cyprus/International Breakpoint Consortium	Scientific Co-ordinator
3	2018-2019	Establishment of an Agricultural Genomics Centre in Cyprus (AGRICYGEN)	H2020-WIDESPREAD-04-2017-TeamingPhase1	Research Team Member
4	2017-2020	Genetic & Molecular Characterisation of Cypriot Patients with Multiple Malformation Syndromes (MMS) of Unknown Aetiology	Telethon Cyprus	Scientific Co-ordinator
5	2020-2023	Fetal genetic factors in pregnancy loss	Telethon Cyprus	Scientific Co-ordinator
6	20203-2026	Mapping breakpoints and identifying cryptic chromosomal rearrangements using next-generation cytogenomic tools.	Telethon Cyprus/Internal Fund	Scientific Co-ordinator
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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	2023-to date	European Molecular Quality Network	Assessor	Assessing laboratories (reports) on specific disorders
2	2021-to date	Cyprus Medically Assisted Reproduction (CMAR) Council	Board Member	Members of CMAR are Assigned by the Council of Ministers to regulate and monitor assisted reproduction in Cyprus
3	2013-2014 and 2016-2018	Cyprus Society of Human Genetics	Board Member	Elected board member
4	2018-2022	Molecular Cytogenetics Journal	Editorial board member	Editorial/Reviewer
5	2023-to date	Frontiers in Genetics	Editorial board member	Editorial board

**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:
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**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	2015	Certified Trainer for Executive Courses	Cyprus Human Resource Development Authority (HRDA) No: E001402
2	2012-2024	Accredited Laboratory Director.	The College of American Pathologist (CAP), USA. LAP No: 6848901
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