

## NICHOLAS K MOSCHONAS (Jan. 2022)

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### A. EDUCATION & PROFESSIONAL TRAINING

Division of Gene Structure and Expression, National Institute of Medical Research (NIMR), Mill Hill, London, Medical Research Council (MRC), UK	Human Molecular Genetics	Postdoctoral Fellow	1979-82
Department of Biology, University of Athens, GR, and Department of Cellular & Developmental Biology, Harvard University, USA	Developmental & Molecular Evolution	Ph. D.	1975-79
Department of Biology, University of Patras, GR	Biology	B. Sc.	1970-75

### B. ACADEMIC AFFILIATIONS

- 2020-present: Affiliated Scientist, FORTH/ICE-HT, GR
- 2020-present: Professor Emeritus of Biology and Medical Molecular Genetics, Medical School, University of Patras, GR, ([n\\_moschonas@med.upatras.gr](mailto:n_moschonas@med.upatras.gr), cell. Phone: 00306977783551)
- 2006-2020: Professor of Biology and Medical Molecular Genetics, Medical School, University of Patras, GR; 2009-2020: Director of the Laboratory of General Biology, Medical School, U Patras, GR
- 2014-2020: Collaborating Professor Member of the Institute of Chemical Engineering Science-FORTH Patras, GR.
- 1998-2006: Professor of Human Molecular Genetics (HMG) and Head of the HMG Laboratory (1984-2006), Department of Biology University of Crete, GR; 2000-2004: Vice-Chairman of the Department. [1993-1988: Associate Professor; 1988-1993: Assistant Professor; 1984-1988: Lecturer, Department of Biology, U. of Crete, GR].
- 1983-2006: Research Scientist (joint appointment), Institute of Molecular Biology and Biotechnology (IMBB), Foundation of Research and Technology (FORTH), Heraklion, Crete, Greece.

### C. EDUCATION & TRAINING

- 1979-1982: EMBO Postdoctoral Fellow, Division of Gene Structure & Expression, Director: Richard A. Flavell, National Institute of Medical Research, Medical Research Council, Mill Hill, London, UK
- 1975-1979: Graduate student, PhD candidate supported by a fellowship of the State Fellowships Foundation (SFF-GR), Greece; Dept. of Biology, U. of Athens and Dept of Cellular & Developmental Biology, the Biological Laboratories, Harvard U., MA, USA (Supervisor: Prof. Fotis C Kafatos).
- 1970-1975: BSc in Biology, Dept. of Biology, U. of Patras, annually awarded by SFF-GR for the highest grade scoring of the class.

### D. RESEARCH INTERESTS & ACADEMIC ACTIVITIES

- **RESEARCH INTERESTS:** Medical molecular genetics and functional genomics with the aim to understand the architecture and the biological impact of the genetic information in health and disease exploiting cell and animal models combined with bioinformatics and integrated omics approaches.
- Coordinator or Principal Investigator of 11 international [NIH US (2), UNIDO grants (1) & EU (8) grants] and 24 national competitive research grants.
- Total number of publications and review articles: 93, Average Impact Factor: 6.2, Citations: 2980, h-index: 27, i10-index:43. More than 70 invited talks in Scientific Congresses and Meetings.
- Co-organizer of 28 international or national scientific symposia (1990-2021).

- More than 245 Short Communications and/or Abstracts in international and national Conference Proceedings Volumes.

➤ **ACADEMIC ACTIVITIES**

- Member of the Human Genome Organization (HUGO), the European and the American Societies of Human Genetics, the International Mammalian Genome Society, the Hellenic Association of Medical Geneticists, the Hellenic Society of Biochemistry and Molec. Biology, The Hellenic Society of Computational Biology and Bioinformatics, Senior Editor of Genome Database (GDB) for Chromosome 10 elected by the Human Genome Organisation/Human Genome Mapping Committee, 1999- 2003.
- Science Management: National Delegate of Greece for the EU FP7 Theme “Cooperation-HEALTH” (2005-2010), Assoc. Member of the National Council for Research & Technology (ESET) of Greece (2005-2009), Member of the *ad hoc* National Committee for the National Strategic Reference Framework (NSRF) for Research, Technical Development & Innovation-GR, National Delegate/ Expert at the Human Genome Analysis Program of the CAN-MED Biomedicine and Health Research Program, EC-DGXII, 1992- 1997, Member of the European Community Working Party for the “Human Genome Analysis” Research Project, (1988-1990).

**E. TEACHING & SCIENTIFIC SUPERVISING**

- (1983-present): Full-semester under- and/or postgraduate-level courses in Medical Molecular Genetics, Genomics and Molecular Cell Biology [Med. School, U. Patras (2006-present), Dept. of Biology, U. of Crete (1984-2006) as faculty member]; AUTH, NKUA, DUTH, U. CRETE (as Invited Instructor, since 2008).
- (1984-present): Academic Supervisor of 13 PhD, 20 MSc theses, and 20 Diploma theses.
- (1990-present): Member of the Advisory and Examination Committees for more than 90 PhD or MS dissertations.

**F. ACADEMIC APPOINTMENTS (RECENT AND INDICATIVE)**

- Chairman of the Basic Medical Sciences I Division, Medical School, U. Patras (2018-2020).
- Member of the General Assembly, Medical School, U. Patras (2016-2020).
- Director of the Dept. of General Biology, (2009-2020).
- National Delegate of Greece at the EU FP7 Theme “Cooperation-HEALTH” (2005-2010).
- Deputy Member of the National Council for Research & Technology-GR (2005-2009).
- Member of the Bioethics Committee of the Hellenic Orthodox Church (2002-present).
- HUGO/Genome Database (GDB) Editor for Human Chromosome 10 (1995-2004); Senior Editor for HC10, elected by the Human Genome Organization/Human Genome Mapping Committee, 1999- 2004.
- Member of the *ad hoc* Committee for the National Strategic Reference Framework (NSRF) for Research, Technical Development & Innovation-GR (Section: Health) (2008-09).
- Deputy Chair of the Department of Biology, University of Crete (2000-04).
- National Expert at the Human Genome Analysis Program of the CAN-MED Biomedicine and Health Research Program, EC-DGXII, 1992- 1997.
- Member of the Research Committee (1999-02), and the Senate of the University of Crete (1988-90).

➤ **SELECTED PUBLICATIONS**

- 1) Moschonas N.K., de Boer E., Grosveld F.G., Dahl H.H.M., Wright S., Shewmaker, C.K., Flavell, R. A (1981). Structure and expression of a cloned  $\beta$ -thalassaemic globin gene. *Nucleic Acids Res.* 9:4391-4401
- 2) Busslinger, M., Moschonas, N. and Flavell, R.A (1981). Beta+ thalassaemia: Aberrant splicing results from a single point mutation in an intron. *Cell* 27:289-298.

- 3) Flavell, R.A., Bud, H., Bullman, H., Dahl, H., de Boer, E., de Lange, T., Groffen, J., Grosveld, F., Grosveld, G., Kioussis, D., Moschonas, N. and Shewmaker, C. (1981). Globin gene expression in vivo and in vitro. In: *Organization and Expression of Globin Genes.* Eds. G. Stamatoyannopoulos and A.W. Nienhuis, **2nd Conference on Hemoglobin Switching**, Airlie, Virginia, U.S.A. Alan R. Liss Inc., New York, pp. 119-126 (review).
- 4) Moschonas, N.K., de Boer, E., Flavell, R.A. (1982). The DNA sequence of the 5' flanking region of the human beta globin gene: evolutionary conservation and polymorphic differences. *Nucleic Acids Res.* 10, 2109-2120.
- 5) Rodakis, G., Moschonas, N. and Kafatos, F.C. (1982). Evolution of a Multigene Family of Chorion Proteins in Silkworms. *Mol. Cell. Biol.*, 2, 554-563.
- 6) Rodakis, G., Moschonas, Regier, J. and Kafatos, F.C. (1983). The B Multigene family of Chorion Proteins in Saturniid Silkworms. *J. Mol. Evol.*, 19, 322-332.
- 7) Moschonas, N.K., Thireos, G. and Kafatos, F.C (1988). Evolution of Chorion Structural Genes and Regulatory Mechanisms in Two Wild Silkworms: A Preliminary Analysis. *J. Mol. Evol.* 27:187-193.
- 8) Mavrothalassitis, G, Tzimagiorgis, G, ..., Plaitakis, A., Papamatheakis, J and Moschonas, N.K (1988). Isolation and characterization of cDNA clones encoding human liver glutamate dehydrogenase: Evidence for a small gene family. *Proc. Natl. Acad. Sci. USA*, 85:3494-3498.
- 9) Mamalaki, A. and Moschonas, N.K. (1990). Aberrance and modification of alpha-1 and alpha-2 globin gene expression in human and mouse erythroleukemia cells. *Acta Haematol.*, 84, 30-37.
- 10) Mamalaki A, Anagnou N., Moschonas N. (1990). Developmental and inducible patterns of human theta-1-globin gene expression in embryonic/fetal and adult erythroid cells. *Am. J. Haematol.* 35:251-257.
- 11) Tzimagiorgis, G., Adamson, M.C., Kozak, C.A. & Moschonas N.K (1991). Chromosomal mapping of glutamate dehydrogenase gene sequences to mouse chromosomes 7 and 14. *Genomics*, 10:83-88.
- 12) Michaelidis Th, Tzimagiorgis G. Moschonas N.K. & Papamatheakis J. (1993). The human glutamate dehydrogenase (GLUD) gene family: gene structure and organization. *Genomics*, 16:150-160.
- 13) Deloukas, P., Dauwerse, J.G., Moschonas, N.K., van Ommen, G.J.B. and van Loon, A.P.G.M. (1993). Three human glutamate dehydrogenase genes (GLUD1, GLUDP2 and GLUDP3) genes are located on chromosome 10q, but are not closely physically linked. *Genomics*, 17, 676-681.
- 14) Moschonas, N.K., ..Lubyova, B., Manifava, M., Deloukas, P., van Loon, G-J.B. and M. Kapsetaki (1993). Dinucleotide repeat polymorphism (D10S608) adjacent to the GLUD1 locus. *Hum. Molec. Genet.* 2, 11, 1981.
- 15) Kapsetaki, M., Kokkinaki, M., Angelicheva, D., Lubyova, B., Mavraki, H., Argyrokastritis, A., Ferguson-Smith, M., Lush, M. and Moschonas, N.K. (1994). The EUROGEM map of human chromosome 10. In, N.K.Spurr et al.: European Gene Mapping Project (EUROGEM): Genetic maps based on the CEPH reference families. *Eur. J. Hum. Genet.* 2,193-252.
- 16) Pucyrivov, A.T., Chroniary, K and Moschonas, N.K. (1995). Normalized cDNA library from human erythroleukemia cells. *Molec. Biol. (Mosc)*, 29, 1, 58-61.
- 17) Anagnou, N.P., Perez-Stable, C., Gelinas, G., Constantini, F., ..Costeas, T., Moschonas, N.K. and Stamatoyannopoulos, G (1995). Sequences located 3' to the brakepoint of the HPFH-3 deletion exhibit enhancer activity and can modify the developmental expression of the human fetal A-gamma-globin gene in transgenic mice. *J. Biol. Chem.*, 270, (17):10256-10263.
- 18) Moschonas, N.K., Spurr, N.K. and Mao, J-I (1996). Report of the First International Workshop on Human Chromosome 10 mapping 1995. *Cytogenet Cell Genet.* 72:99-112.
- 19) Kritis, A.A., Argyrokastritis, A., Moschonas, N.K., Power, S., Katrakili, N., Zannis, V.I., Cereghini, S. and Talianidis, I (1996). Isolation and characterisation of a novel isoform for the human hepatocyte nuclear factor 4. *Gene* 173:275-280.
- 20) Cox, S.A., ..Povey, S., Rebello, M., Kapsetaki, M., Moschonas, N.K., Grzeschik, K.-H., Otto, M., Dixon, M., .. F., Wright, A., Teague, P., Terrenato, L., Gal, A., Mueller-Myhsok, B., Cann, H.M. and Spurr, N.K. (1996). European Gene Mapping Project (EUROGEM): Breakpoint panels for human chromosomes based on the CEPH reference families. *Ann. Hum. Genet.* 60:447-486.

- 21) Liu, D., Pavlopoulos, E., ..Moschonas, N. and Mavrothalassitis, G (1997). ERF: Genomic organization, chromosomal localization and promoter analysis of the human and the mouse genes. *Oncogene* 14:1445-1451.
- 22) Shashidharan, P., ..Moschonas, N.K. and Plaitakis, A. (1997). Nerve tissue-specific human glutamate dehydrogenase that is thermolabile and highly regulated by ADP. *J. Neurochem.* 68:1804-1811.
- 23) Marzella, R., Kokkinaki, M.A., Kapsetaki, M., Ricco, A., Argyrokastritis, A., Kamakari, S., Sarafidou, T., Archidiacono, N., Roussou, A., Paspadaki, A., Rocchi, M. and Moschonas, N.K. (1997). Map integration at human chromosome 10: molecular and cytogenetic analysis of a chromosome-specific somatic cell hybrid panel and genomic clones, based on a well-supported genetic map. *Cytogenet. Cell Genet.* 79:257-265
- 24) Mavrogiannis L, Argyrokastritis A, ..Dermitzakis E, Sarafidou T, Patsalis PC, and Moschonas NK (2001). ZNF232: Structure and expression analysis of a novel human C2H2 zinc finger gene, member of the SCAN/LeR domain subfamily. *Biochem. Biophys. Acta*, Apr 16;1518(3):300-305
- 25) Bentley, D.R, Deloukas P, ... Moschonas N.K, ... Sarafidou T, .. et al., (2001). The Physical Maps for Sequencing Human Chromosomes 1, 6, 9, 10, 13, 20 and X. *Nature*, **409**, 942 – 943.
- 26) Pavlopoulos E., Pitsouli C, Klueg K, Moschonas NK, and Delidakis, C. (2001). *neuralised* encodes a peripheral membrane protein involved in Delta signalling and endocytosis. *Develop Cell*, 1, 807-816
- 27) Morante-Redolat JM, ..Gesck S., Sarafidou T., Mautner V-F, ...Deloukas P, Moschonas N K, Michelucci R, Siebert R, Nobile C, Pérez-Tur J, López de Munain A. (2002). Mutations in the LGI1/Epitempin gene on 10q24 cause autosomal dominant lateral temporal epilepsy. *Human Molec Genet.* 11(9), 1119-1128.
- 28) Staub E., Perez-Tur J., Siebert R., Nobile C., Moschonas N.K., Deloukas P. and Hinzmann B. (2002). The novel EPTP repeat defines a superfamily of proteins with implications in epileptic disorders. *Trends Biochem Sci.* 27 (9) 441-444
- 29) Sarafidou T, . . . Baker E, Kokkinaki M, ... Deloukas P, Sutherland G R, Kutche K, Moschonas N K, Siebert R, Gecz J. (2004). Folate-sensitive fragile site FRA10A is due to an expansion of a CGG-repeat in a novel gene FRA10AC1, encoding a nuclear protein. *Genomics* 84(1): 69-81
- 30) Deloukas P., Earthrowl M.E..... Kokkinaki, M.,.....Sarafidou T., Sehra H.K.,.....Lovering, R.C., Moschonas NK., Siebert R., Fechtel K., Bentley D., ...Smith DR., and Rogers J. (2004). The DNA sequence and comparative analysis of human chromosome 10. *Nature*, 429, 375 – 381
- 31) Mizi A, Zouros E, Moschonas N & Rodakis GC (2005). The complete maternal and paternal mitochondrial genomes of the Mediterranean mussel *Mytilus galloprovincialis*: Implications for the Doubly Uniparental Inheritance mode of mtDNA. *Mol Biol Evol* 22(4): 952-967
- 32) Kartsaki E, Spanaki C, .. Moschonas N, Macdonald M, Plaitakis A. (2006). Late-onset and typical Huntington disease families from Crete have distinct genetic origins. *Int J Mol Med.* 17(2):335-46.
- 33) Ayerdi-Izquierdo, A, Stavrides, G., .....Sarafidou, T., Hinzmann, B., Moschonas, N., Siebert, R., Deloukas, P., Nobile, C., Pérez-Tur, J. (2006). Genetic analysis of the LGI/Epitempin gene family in sporadic and familial lateral temporal lobe epilepsy. *Epil Res* Aug;70(2-3):118-26.
- 34) Martinez L, Underhill P, .., Moschonas N, ..Cavalli-Sforza L, Herrera RJ. (2007). Paleolithic Y-haplogroup heritage predominates in a Cretan highland plateau. *Eur. J. Hum Genet.* **15**, 485–493
- 35) Athanasiadis G, ...Moschonas N, Chaabani H, Moral P (2007). The X chromosome Alu insertions as a tool for human population genetics: data from European and African human groups. *Eur. J. Hum Genet.* **15**, 578–583.
- 36) Koutelou, E, Sato S, ..Kokkinaki M, Conaway RC, Conaway J W, and Moschonas N K. (2007). Neuralized-like 1 (Neurl1) Targeted to the Plasma Membrane by N-Myristoylation Regulates the Notch Ligand Jagged1. *J Biol. Chem.* 283, 3846-3853
- 37) Athanasiadis G, Esteban E, .. Moschonas N, . Moral P. (2010). Different evolutionary histories of the coagulation factor VII gene in human populations? *Ann Hum Genet.* Jan;74(1):34-45.
- 38) Sarri C, Douzgou S, . Sarafidou T, ..Moschonas NK, Petersen MB (2011). Complex distal 10q rearrangement in a girl with mild intellectual disability: Follow up of the patient and review of the literature of non-acrocentric satellite chromosomes. *Am J Med Genet* Part A 155:2841–2854.

- 39) Klapa, MI, Tsafou, K., Theodoridis E., Tsakalidis A. and Moschonas NK (2013). Reconstruction of the experimentally supported human protein interactome: what can we learn? *BMC Syst Biol.* 2013 Oct 2;7(1):96.
- 40) Sarafidou T and Moschonas N (2017). Chromosome 10. *Encyclopedia of Life Sciences (eLS)*, Edit. J. Wiley & Sons. Ltd: Chichester, DOI: 10.1002/9780470015902.a0005819.pub3, pp:1-43.
- 41) van Rijswijk M, ..Klapa M I, ..Le Corguillé G, Moschonas N K, ..Reczko M, ..Steinbeck C. (2017). The future of metabolomics in ELIXIR. *F1000Research* 2017, 6 (ELIXIR):1649 Last updated: 18 Sep 2017.
- 42) Gioutlakis A, Klapa MI, and Moschonas NK. (2017). PICKLE 2.0: A human protein-protein interaction meta-database employing data integration via genetic information ontology. *PLoS ONE* PONE-D-17-20180R1; DOI: 10.1371/journal.pone.0186039.
- 43) Papadimitropoulos M, Anastasopoulou S, Galiopoulou E, Manousopoulou A, Bicciato S, Garbis S, Sarafidou Th. Klapa MI, and Moschonas N.K. (2018). Integrated high-throughput biomolecular analyses of *FRA10AC* altered expression in a human cell model. *Eur J Hum Genet*: 50<sup>th</sup> Eur. Society of Human Genetics Conf., <https://doi.org/10.1038/s41431-018-0247-7>, P16.32D.
- 44) E. Tsare, A. Gioutlakis, M. I. Klapa, N. K. Moschonas (2019). Investigating the genetic architecture of hypertension through combined analysis of genome-wide association studies (GWAS) data and the human protein interaction network. *Eur J Hum Genet* 27, 1–688 (2019). 51<sup>st</sup> European Society of Human Genetics Conference. <https://doi.org/10.1038/s41431-019-0404-7>; P16.39C.
- 45) GN Dimitrakopoulos, A Gioutlakis, MI Klapa, NK Moschonas (2020) Evaluating the expansion of the experimentally determined human protein interactome using the PICKLE meta-database *Eur J Hum Genet* 27: 1705.
- 46) GN Dimitrakopoulos, MI Klapa, NK Moschonas (2021). PICKLE 3.0: enriching the human meta-database with the mouse protein interactome extended *via* mouse–human orthology. *Bioinformatics*, Volume 37, Issue 1, 1 January 2021, pages 145–146.
- 47) GN Dimitrakopoulos, MI Klapa, NK Moschonas (2022). How Far Are We from the Completion of the Human Protein Interactome Reconstruction? *Biomolecules* 2022, 12(1), 140

For the full publication list see: <https://www.ncbi.nlm.nih.gov/pubmed/?term=Moschonas+N>, & [https://scholar.google.com/citations?user=8L\\_F\\_n4AAAAJ](https://scholar.google.com/citations?user=8L_F_n4AAAAJ).