CURRICULUM VITAE

Kleopas A. KLEOPA, MD, PhD, FAAN, FEAN

Date and place of Birth: May 9, 1968 - Nicosia, Cyprus
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CURRENT POSITION

Professor and Senior Consultant Neurologist, The Cyprus Institute of Neurology & Genetics

- Head of Neuroscience Department
- Coordinator, Center for Neuromuscular Disorders
- Coordinator, Clinical Sector
- Coordinator or MSc/PhD Neuroscience Program
- Head of Neuropathology Laboratory

EDUCATION

1987-1993 Medical Degree, University of Würzburg Medical School, Germany
1991-1994 Doctoral Thesis Project in the Laboratory of Clinical Neurochemistry and Doctor of Medicine Award (grade: "*magna cum laude*"), Medical Faculty of the University of Würzburg

MEDICAL TRAINING

1994-1995 Internship in Neurology, Neurological Clinic of the University, Würzburg, Germany
1995-1996 Internship in Internal Medicine, Medical College of Pennsylvania, Philadelphia, USA
1996-1999 Resident in Neurology, Medical College of Pennsylvania and Hahnemann/Drexel University, Philadelphia
1999 Chief Resident in Neurology, Medical College of Pennsylvania and Hahnemann/Drexel University, Philadelphia
1999-2001 Clinical Fellowship in EMG and Neuromuscular Disorders, University of Pennsylvania Medical Center, Philadelphia
2000-2002 Postdoctoral Research Fellowship, Funded by National Multiple Sclerosis Society, University of Pennsylvania
1999-2001 Muscular Dystrophy Association (MDA) Clinic, Children's Hospital of Philadelphia
2000-2001 Patient Oriented Research Training Program, University of Pennsylvania, Clinical Research Center

POSITIONS HELD AND ACADEMIC EXPERIENCE

2001-2002 Clinical Instructor of Neurology, University of Pennsylvania Medical Center, Philadelphia, USA
2002- Senior Consultant Neurologist, Cyprus Institute of Neurology and Genetics

2007- Head of Neuroscience Department, Cyprus Institute of Neurology and Genetics (CING)
2012- Professor, Cyprus School of Molecular Medicine/CING
2015- Coordinator, Neuroscience Postgraduate Program CING
2020- Coordinator, Center for Neuromuscular Disorders, CING
2024- Head of Neuropathology Laboratory, CING
2021-2024 Coordinator for Research, The Cyprus Institute of Neurology and Genetics

Professional licenses and specialist board certification

1995 License to practice medicine in Germany
1999 Medical License for the State of Pennsylvania, USA
2000 Registration as a Medical Doctor in Cyprus (CMA Nr: 3507)
2000 Recognition as a Specialist Neurologist, Ministry of Health, Cyprus
2002 Diplomate, American Board of Psychiatry and Neurology, Inc.

Languages spoken

Greek (mother language), English, German (professionally fluent)

HONORS AND AWARDS

1987-89 Scholarship of the Cyprus Government to attend Medical School
1990-93 Scholarship of the German Academic Exchange Service (DAAD)
1990 Participation in Erasmus Program for medical students with distinction in Neuroscience, Leiden, the Netherlands
1999 Neurology Teaching Award from Medical Students, Class of 1999, MCP-Hahnemann University
2000-03 Postdoctoral Fellowship Award, National Multiple Sclerosis Society USA
2000-02 Development Grant Award, USA Muscular Dystrophy Association
2009 Fellowship Award, International Peripheral Nerve Society 2009 Meeting
2016-19 Member of the European Science Foundation (ESF) College of Expert Reviewers
2015 **European Academy of Neurology Investigator Award 2015,** Scientific Subspecialty Panel on Neuropathies
2017 **Cyprus National Distinguished Researcher Award, Research Promotion Foundation**
2019 Elected Fellow of the American Academy of Neurology
2021 Elected Fellow of the European Academy of Neurology
2025 Research Commercialization Award (team), Research and Innovation Agency

PROFESSIONAL MEMBERSHIPS AND OTHER ACADEMIC ACTIVITIES

2002 Diplomate, American Board of Psychiatry and Neurology, Inc
1996- Member of the American Academy of Neurology
2000- Member of the American Association of Neuromuscular and Electrodiagnostic Medicine
2002- Member of the Cyprus Neurological Society
2003- Member of the Society for Neuroscience (USA)
2003- Founding Member of the Cyprus Society of Human Genetics
2004-14 Member of the Board of the Cyprus Neurological Society
2005- Member of the World Muscle Society

2005- Scientific Advisor, Cyprus Myasthenia Gravis Association
2005- Member of the European Academy of Neurology
2008- Member of the Peripheral Nerve Society
2010- Member of the International Society of Neuroimmunology
2006-09 President of the Cyprus Bioethics Review Committee for Biomedical Research
2010-14 Cyprus representative in the EU Medical Specialties (UEMS)-Section of Neurology
2011-13 Cyprus representative in the EU Program Committee Health for FP7
2011-13 Cyprus representative in the EU Innovative Medicines Initiative (IMI)
2015-23 Member of the Cyprus National Bioethics Committee
2018- Member of Scientific Advisory Board, Charcot-Marie-Tooth Association (CMTA)
2019- Member of the Scientific Advisory Board, Gene Therapy Initiative for Neurofibromatosis-1, Gilbert Family Foundation, USA
2019- Board Member of the International Charcot-Marie-Tooth and Related Disorders (CMTR) Special Interest Group/Peripheral Nerve Society (*re-elected 2023*)
2019- Fellow of the American Academy of Neurology (FAAN)
2021-23 Member of the Education Committee, Peripheral Nerve Society
2021-24 Member, Scientific Program Committee, Peripheral Nerve Society Meeting
2021- Elected Fellow of the European Academy of Neurology (FEAN)
2022- Member of the American Society for Cell and Gene Therapy (ASCGT)
2022-23 Co-Chair, 2023 Scientific Program Committee, Peripheral Nerve Society Meeting
2022- Cyprus representative in the European Partnership for Rare Disorders (EP-RD)
2023- Member of the Musculoskeletal Disorders Committee, American Society for Gene and Cell Therapy (ASGCT)
2022-24 Member of the Scientific Program Committee, ASGCT
2023- Chair, Inter-ERN (European Reference Networks) Working Group for Gene Therapy
2024- co-Chair, Working Group Leukodystrophies European Reference Network for Rare Neurological Disorders (ERN-RND)
2024- co-Chair, Working Group Neuropathies, European Reference Network for Rare Neuromuscular Disorders (ERN-EURO-NMD)
2024- Member of the Scientific Advisory Board, Pelizaeus-Merzbacher Disease (PMD) Foundation, USA
2024-24 Member of the Research Advisory Committee, CMT Research Foundation (CMTRF)
2024- Founding Member, European CMT Research Association (ECRA)

Reviewer for scientific journals (2000-today): Journal of Neuroscience, Acta Neuropathologica, Muscle and Nerve, Journal of Neurology, Brain, BioMed Central Neurology, Biotechnology Journal, Journal of Inherited Metabolic Disease, Neuron Glia Biology, Acta Myologica, Journal of the Peripheral Nervous System, PLoS Genetics, Journal of Neuropathology and Experimental Neurology, Neurobiology of Disease, Neuromolecular Medicine, PLos One, Glia, Gene Therapy, Frontiers in Molecular Neuroscience, Scientific Reports, Nature Communications, Neurotherapeutics, Experimental Neurology, Journal of Clinical Investigation, Molecular Therapy, EBioMedine, and others.

Reviewer for Research Funding Organizations (2006-today): National Multiple Sclerosis Society, USA, The Wellcome Trust, UK, Association Française contre les Myopathies-AFM, France, ARSEP, France, European Science Foundation (ESF), French National Research Agency, European Leukodystrophy Association (ELA), France, Vaincre les Maladies

Lysosomales (VML), France, Hellenic Foundation for Research and Innovation, MS Society Australia, Prinses Beatrix Spierfonds Netherlands, Auckland Medical Research Foundation, New Zealand, Review Panel Expert for «Medical and Health Sciences» of the European Co-operation in the Field of Scientific and Technical Research - COST Association, European Commission 2015-19 MSCA proposals, 2016 Personalized Medicine Call evaluation, invited expert, COST-2016-2019 Review Panel

Editorial Board Memberships: 2018- Frontiers in Neuroscience; 2019- International Journal of Molecular Sciences; 2022- Neuroimmunology (Greece), 2022- Journal of the Peripheral Nervous System

REVIEW OF SCIENTIFIC WORK and CONTRIBUTION TO SCIENCE

Work outlined below was selected from a career total of 130 full papers, 20 book chapters and over 130 oral or poster presentations (more than 8600 citations in total to date, h-index=42) – last updated March 2025. Full list at:
http://scholar.google.com/citations?user=uqquFR4AAAAJ&hl=en
http://www.ncbi.nlm.nih.gov/pubmed/?term=kleopa

1. Disease mechanisms in inherited neuropathies: Mainly focusing on X-linked Charcot-Marie-Tooth Disease (CMT1X), but also on other CMT types including CMT2A, CMT1A, CMT4C, my research has contributed to the understanding of demyelination and axonal loss mechanisms, as well as of the repertoire of Schwann cell and oligodendroglial gap junction protein expression, their anatomic and functional relationship and possible interactions in health and disease. I contributed the discovery and characterization of Cx29 and Cx47 in glial cells, and I generated novel mouse models of CMT1X and CMT2A, clarifying the cellular mechanisms involved. I also contributed to the establishment of responsive blood biomarkers. *Related publications*:

1) **Kleopa KA**, Yum SW, Scherer SS (2002) Cellular mechanisms of connexin32 mutations associated with CNS manifestations. *J. Neurosci. Res.* 68:522-534.
2) Altevogt BM*, **Kleopa KA***, Postma FR, Scherer SS, Paul DL (2002) Connexin29 Is Uniquely Distributed within Myelinating Glial Cells of the Central and Peripheral Nervous Systems *J. Neurosci.,* 22: 6458-6470. (*equal contribution)
3) **Kleopa KA**, Orthmann JL, Enriquez A, Paul DL, Scherer SS (2004) Unique distribution of gap junction proteins connexin29, connexin32, and connexin47 in oligodendrocytes, *Glia,* 47:346-57.
4) Sargiannidou I, Vavlitou N, Aristodemou S, Hadjisavvas A, Kyriacou K, Scherer SS, **Kleopa KA** (2009). Connexin32 mutations cause loss of function in Schwann cells and oligodendrocytes leading to PNS and CNS myelination defects. *J Neurosci*, 29:4748-4761.
5) Sargiannidou I, Kim GH, Kyriakoudi S, Eun BL, **Kleopa KA.** (2015) A start codon CMT1X mutation associated with transient encephalomyelitis causes complete loss of Cx32. *Neurogenetics*, 16:193-200.
6) Olympiou M, Sargiannidou I, Markoullis K, Karaiskos C, Kagiava A, Kyriakoudi S, Abrams CK, **Kleopa KA** (2016). Systemic inflammation disrupts oligodendrocyte gap junctions and induces ER stress in a model of CNS manifestations of X-linked Charcot-Marie-Tooth disease. *Acta Neuropathol Commun*. Sep 1;4(1):95.
7) Kyriakoudi S, Sargiannidou I, Kagiava A, Olympiou M, **Kleopa KA** (2017) Golgi-retained Cx32 mutants interfere with gene addition therapy for CMT1X. *Hum Mol Genet*. 2017 26:1622-163.

8) Stavrou M, Sargiannidou I, Christofi T, **Kleopa KA**. Genetic mechanisms of peripheral nerve disease. *Neurosci Lett*. 2020 Nov 26:135357. doi: 10.1016/j.neulet.2020.135357.
9) Stavropoulos F, Sargiannidou I, Potamiti L, Kagiava A, Panayiotidis MI, Bae JH, Yeom SC, Lee JY, **Kleopa KA**. Aberrant Mitochondrial Dynamics and Exacerbated Response to Neuroinflammation in a Novel Mouse Model of CMT2A. *Int J Mol Sci.* 2021 Oct 26;22(21):11569. doi: 10.3390/ijms222111569.PMID: 34769001
10) Jennings MJ, Kagiava A, Vendredy L, Spaulding EL, Stavrou M, Hathazi D, Grüneboom A, De Winter V, Gess B, Schara U, Pogoryelova O, Lochmüller H, Borchers CH, Roos A, Burgess RW, Timmerman V**, Kleopa KA**, Horvath R. (2022) NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. ***Brain***. 2022; 145:3999-4015. doi: 10.1093/brain/awac055.
11) Stavropoulos F, Georgiou E, Schiza N, Bell S, Baloh RH, **Kleopa KA**, Sargiannidou I. Mitofusin 1 overexpression rescues the abnormal mitochondrial dynamics caused by the Mitofusin 2 K357T mutation in vitro. *J Peripher Nerv* Syst. 2023; 28:329-340. doi: 10.1111/jns.12564.

2. Gene Therapy Development: In addition to clarification of the molecular mechanisms of inherited neuropathies and leukodystrophies, and based on results from this work, we developed in the last 15 years novel gene therapy approaches to replace or silence neuropathy-associated genes specifically in myelinating glial cells of the peripheral and central nervous systems. Using relevant models of neuropathy and leukodystrophy, we have explored world-wide innovative gene therapy approaches using lentiviral and AAV vectors for cell-targeted expression based on cell-specific promoters. *Related publications*:

1) Kagiava A, Sargiannidou I, Bashiardes S, Richter J, Schiza N, Christodoulou C, Gritti A, **Kleopa KA**. (2014) Gene delivery targeted to oligodendrocytes using a lentiviral vector. *J Gene Med*. 16(11-12):364-73.
2) Schiza N., Sargiannidou I., Kagiava A., Karaiskos C., Nearchou M., **Kleopa KA** (2015) Transgenic replacement of Cx32 in gap junction deficient oligodendrocytes rescues the phenotype of a hypomyelinating leukodystrophy model. *Hum Mol Genet*, 24: 2049-64.
3) Sargiannidou I, Kagiava A, Bashiardes S, Richter J, Christodoulou C, Scherer SS, **Kleopa KA** (2015) Intraneural GJB1 gene delivery improves nerve pathology in a model of CMT1X. *Annals of Neurology*, 78:303-316.
4) Kagiava A, Sargiannidou I, Theophilidis G, Karaiskos C, Richter J, Bashiardes S, Schiza N, Nearchou M, Christodoulou C, Scherer SS**, Kleopa KA** (2016) Intrathecal gene therapy rescues a model of demyelinating peripheral neuropathy. *Proc Natl Acad Sci U S A*, 113 (17):e2421-9.
5) Georgiou E, Sidiropoulou K, Richter J, Papaneophytou C, Sargiannidou I, Kagiava A, von Jonquieres G, Christodoulou C, Klugmann M, **Kleopa KA** (2017) Gene therapy targeting oligodendrocytes provides therapeutic benefit in a leukodystrophy model, *Brain,* 140:599-616.
6) Kagiava A, Karaiskos C, Richter J, Tryfonos C, Lapathitis G, Sargiannidou I, Christodoulou C, **Kleopa KA** (2018). Intrathecal gene therapy in mouse models expressing CMT1X mutations. *Hum Mol Genet.* 27: 1460-1473.
7) Schiza N, Georgiou E, Kagiava A, Médard J-J, Richter J, Tryfonos C, Sargiannidou I, Heslegrave AJ, Rossor AM, Zetterberg H, Reilly MM, Christodoulou C, Chrast R, **Kleopa KA** (2019). Gene replacement therapy in a model of Charcot-Marie-Tooth 4C neuropathy, *Brain.* 2019 May 1;142(5):1227-1241.
8) Kagiava A, Richter J, Tryfonos C, Karaiskos C, Heslegrave AJ, Sargiannidou I, Rossor AM, Zetterberg H, Reilly MM, Christodoulou C, **Kleopa KA**. Gene replacement therapy after neuropathy onset provides therapeutic benefit in a model of CMT1X. *Hum Mol Genet.* 2019, 28: 3528-3542.
9) Kagiava A, Karaiskos C, Richter J, Tryfonos C, Jennings MJ, Heslegrave AJ, Sargiannidou I, Stavrou M, Zetterberg H, Reilly MM, Christodoulou C, Horvath R, **Kleopa KA**. AAV9-mediated Schwann

cell-targeted gene therapy rescues a model of demyelinating neuropathy. *Gene Ther*. 2021, doi: 10.1038/s41434-021-00250-0.

10) Stavrou M, Sargiannidou I, Georgiou E, Kagiava A, **Kleopa KA.** Emerging Therapies for Charcot-Marie-Tooth Inherited Neuropathies. *Int J Mol Sci.* 2021 Jun 3;22(11):6048. doi: 10.3390/ijms22116048.
11) Kagiava A, Richter J, Tryfonos C, Leal-Julià M, Sargiannidou I, Christodoulou C, Bosch A, **Kleopa KA**. Efficacy of AAV serotypes to target Schwann cells after intrathecal and intravenous delivery. *Scientific Reports*. 2021 Dec 2;11(1):23358. doi: 10.1038/s41598-021-02694-1.
12) Stavrou M, Kagiava A, Choudury SG, Jennings MJ, Wallace LM, Fowler AM, Heslegrave A, Richter J, Tryfonos C, Christodoulou C, Zetterberg H, Horvath R, Harper SQ, **Kleopa KA.** A translatable RNAi-driven gene therapy silences PMP22/Pmp22 genes and improves neuropathy in CMT1A mice. *J Clin Invest.* 2022 May 17:e159814. doi: 10.1172/JCI159814.
13) Georgiou E, Kagiava A, Sargiannidou I, Schiza N, Stavrou M, Richter J, Tryfonos C, Heslegrave A, Zetterberg H, Christodoulou C, **Kleopa KA**. AAV9-mediated SH3TC2 gene replacement therapy targeted to Schwann cells for the treatment of CMT4C. *Molecular Therapy* 2023, 31(11):3290-3307. DOI: https://doi.org/10.1016/j.ymthe.2023.08.020.
14) Kagiava A, Karaiskos C, Lapathitis G, Heslegrave A, Zetterberg H, Bosch, A, **Kleopa KA.** Gene replacement therapy in two Golgi-retained CMT1X mutants before and after the onset of demyelinating neuropathy. *Molecular Therapy-Meth & Clin Dev*, 2023, 30:377-393.
15) Stavrou M, **Kleopa KA**. Gene therapies for CMT neuropathies: from the bench to the clinic. *Curr Opin Neurol.* 2024 Oct 1;37(5):445-454. doi: 10.1097/WCO.0000000000001289.
16) Christou M, Sargiannidou I, Papacharalambous R, Richter J, Tryfonos C, Christodoulou C, Kagiava A, **Kleopa KA**. A dose escalation and safety study of AAVrh10-mediated Schwann cell-targeted gene therapy for CMT1X. Neurotherapeutics. 2025 Mar 6:e00568. doi: 10.1016/j.neurot.2025.e00568.

3. Glial connexin pathology in acquired demyelination and neurodegeneration: A further line of research in my lab has been the study of gap junctions in multiple sclerosis (MS) post-mortem human brain samples as well as in experimental encephalomyelitis (EAE) and Alzheimer's Disease (5xFAD) mouse models. Detailed analysis of disease brain samples has shown widespread glial connexin pathology with loss of gap junctions in oligodendrocytes not only in MS white and gray matter lesions, but also in normal appearing tissue and in parallel development of astrogliosis and disconnection of astrocytes from oligodendrocytes. These changes correlate with inflammation and disease progression. *Related publications*:

1) Markoullis K, Sargiannidou I, Gardner C, Hadjisavvas A, Reynolds R, **Kleopa KA** (2012) Disruption of oligodendrocyte gap junctions in experimental autoimmune encephalomyelitis. *Glia*, 60:1053-66.
2) Markoullis K, Sargiannidou I, Schiza N, Hadjisavvas A, Roncaroli F, Reynolds R, **Kleopa KA** (2012) Gap junction pathology in multiple sclerosis lesions and in normal appearing white matter. *Acta Neuropathol*, 123:873-86.
3) Markoullis K, Sargiannidou I, Schiza N, Roncaroli F, Reynolds R, **Kleopa KA** (2014) Oligodendrocyte gap junction loss and disconnection from reactive astrocytes in multiple sclerosis grey matter. *J Neuropathol Exp Neurol*, 73(9):865-79.
4) Papaneophytou CP, Georgiou E, Karaiskos C, Sargiannidou I, Markoullis K, Mona Freidin M, Abrams CK, **Kleopa KA** (2018) Regulatory role of oligodendrocyte gap junctions in inflammatory demyelination. *Glia,* 66(12):2589-2603.
5) Papaneophytou C, Georgiou E, **Kleopa KA**. The role of oligodendrocyte gap junctions in neuroinflammation. *Channels* (Austin). 2019 Dec;13(1):247-263. doi: 10.1080/19336950.2019.1631107.

6) Angeli S, Kousiappa I, Stavrou M, Sargiannidou I, Georgiou E, Papacostas SS, **Kleopa KA**. Altered Expression of Glial Gap Junction Proteins Cx43, Cx30, and Cx47 in the 5XFAD Model of Alzheimer's Disease. *Front Neurosci*. 2020; 14: 582934.
7) Stavropoulos F, Georgiou E, Sargiannidou I, **Kleopa KA**. Dysregulation of Blood-Brain Barrier and Exacerbated Inflammatory Response in Cx47-Deficient Mice after Induction of EAE. *Pharmaceuticals* (Basel). 2021 Jun 28;14(7):621. doi: 10.3390/ph14070621.
8) Pechlivanidou M, Kousiappa I, Angeli S, Sargiannidou I, Koupparis AM, Papacostas SS, **Kleopa KA**. Glial Gap Junction Pathology in the Spinal Cord of the 5xFAD Mouse Model of Early-Onset Alzheimer's Disease. *Int J Mol Sci* 2022 Dec 9;23(24):15597. doi: 10.3390/ijms232415597
9) Theophanous S, Sargiannidou I, **Kleopa KA.** Glial Cells as Key Regulators in Neuroinflammatory Mechanisms Associated with Multiple Sclerosis. *Int J Mol Sci*. 2024 Sep 4;25(17):9588. doi: 10.3390/ijms25179588.

4. Autoimmune and paraneoplastic disorders presenting with neuromyotonia and encephalitis: My research has also contributed to the understanding of autoimmune neuropathies focusing on immune-mediated alterations of axonal potassium channel complex and associated molecules. We have contributed to the identification of the antigenic targets and phenotypes involved. *Related publications*:

1) **Kleopa KA**, Elman L, Lang B, Vincent A, Scherer SS (2006). Neuromyotonia and limbic encephalitis sera target mature *Shaker*-type K+ channels: subunit specificity correlates with clinical manifestations. *Brain,* 129:1570-84.
2) Vincent A, Lang B, **Kleopa KA** (2006). Autoimmune channelopathies and related neurological disorders. *Neuron*, 52:123-138.
3) Irani SR, Alexander S, Waters P, **Kleopa K**A, Pettingill P, Zuliani L, Peles E, Buckley C, Lang B, Vincent A (2010) Antibodies to Kv1 potassium channel-complex proteins leucine-rich, glioma inactivated 1 protein and contactin-2-associated protein in limbic encephalitis, Morvan's syndrome and acquired neuromyotonia*. Brain*, 133:2734-2748.
4) Irani SR,* Pettingill P,* **Kleopa KA,*** Schiza N, Waters P, Mazia C, Zuliani L, Watanabe O, Lang B, Buckley C, Vincent A (2012) Morvan's syndrome: clinical and serological observations in 29 cases. *Annals of Neurology*, 72:241-55 (*:*joint first authors*)

5. Epidemiology, diagnosis, genetics and treatment of rare neurological and especially neuromuscular disorders: I have contributed to several research projects focusing on rare neurological and neuromuscular disorders including myasthenia, ALS and peripheral neuropathies, as evident in my numerous publications, among which:

1) Zisimopoulou P, et al. (2014) A comprehensive analysis of the epidemiology and clinical characteristics of anti-LRP4 in myasthenia gravis. J Autoimmun. 52:139-45.
2) Stergiou C, et al. (2016) Titin antibodies in "seronegative" myasthenia gravis - A new role for an old antigen. *J Neuroimmunol*. 2016 Mar 15;292:108-15.
3) Topaloudi A, et al. Myasthenia gravis genome-wide association study implicates AGRN as a risk locus. *J Med Genet*. 2021 Aug 16:jmedgenet-2021-107953. doi: 10.1136/jmedgenet-2021-107953.
4) Koutsis G, et al. Variant transthyretin amyloidosis (ATTRv) polyneuropathy in Greece: a broad overview with a focus on non-endemic unexplored regions of the country. *Neuromuscul Disord.* 2021 Sep 29:S0960-8966(21)00654-4. doi: 10.1016/j.nmd.2021.09.008. Online ahead of print. PMID: 34740514a.
5) Adams D, et al.: Efficacy and safety of vutrisiran for patients with hereditary transthyretin-mediated amyloidosis with polyneuropathy: a randomized clinical trial. HELIOS-A Collaborators. *Amyloid.* 2022 Jul 23:1-9. doi: 10.1080/13506129.2022.2091985.

6) Topaloudi A, et al. PheWAS and cross-disorder analysis reveal genetic architecture, pleiotropic loci and phenotypic correlations across 11 autoimmune disorders. *Front Immunol.* 2023 Sep 21;14:1147573. doi: 10.3389/fimmu.2023.1147573.
7) Votsi C, et al. *RFC1* Repeat Distribution in the Cypriot Population: Study of a Large Cohort of Patients With Undiagnosed Ataxia and Non-Disease Controls. *Neurol Genet*. 2024 Apr 25;10(3):e200149. doi: 10.1212/NXG.0000000000200149.
8) Adams D, et al. Patisiran Global OLE study group. Five-Year Results With Patisiran for Hereditary Transthyretin Amyloidosis With Polyneuropathy: A Randomized Clinical Trial With Open-Label Extension. JAMA Neurol. 2025 Jan 13. doi: 10.1001/jamaneurol.2024.4631. Online ahead of print.

PARTICIPATION IN CLINICAL TRIALS:

- Principal investigator (PI): In Open Label Extension trial OLE ALN-TTR02-006 for familial amyloid neuropathy treatments (2020-23) for ATTRv neuropathy treatment
- Sub-I: In Phase 3 study ALN-TTRSC02-002 (2019-) for ATTRv neuropathy treatment
- Sub-i: In Phase 3 study ION-682884-CS3 (2020-) for ATTRv neuropathy treatment
- PI: In Phase 2/3 study of ARGX-113-2007 v2.0 Alkivia (2022-) for the treatment of inflammatory myopathy
- PI: Phase 3, Open-Label Extension study study of ARGX-113-2011 Alkivia+ (2024-)
- PI: Phase 3 study ARGX-113-2308 (2024-) treatment of seronegative myasthenia
- PI: Phase 3 study ARGX-113-2315 (2025-) for the treatment of ocular myasthenia

PATENTS FILED

- Co-Inventor in the patent WO2020245169A1 “AAV vectors with myelin protein zero promoter and uses thereof for treating Schwann cell-associated diseases like Charcot-Marie-Tooth disease (filed June 2019)
- Co-Inventor in the patent application US63/120,190 “PRODUCTS AND METHODS FOR INHIBITION OF EXPRESSION OF PERIPHERAL MYELIN PROTEIN-22” (filed Dec 2020)

RESEARCH GRANTS OBTAINED (*selection*)

2000-02 Advanced Postdoctoral Fellowship Award (FA 1393-A-1), National Multiple Sclerosis Society USA: Connexin32 mutations and central demyelination ($ 87,319)
2002-04 Advanced Postdoctoral Fellowship Award (FA 1393-A-1), National Multiple Sclerosis Society USA: Connexin32 mutations and central demyelination ($ 87,319)
2004-09 Research Grant (RG 3457A2/1) National Multiple Sclerosis Society USA “CNS connexins and demyelination in CMTX” ($ 219,750)-PI
2007-09 Research Grant of the Research Promotion Foundation: “Models of demyelinating neuropathy and encephalopathy” (90,000 CYP)-PI
2008-10 Research Grant of the Research Promotion Foundation: “The role of gap junctions in Multiple Sclerosis” (120,000 Euro)-PI
2010-12 Research Grant, Research Promotion Foundation (Access to Infrastructure Grant): “Gap junction pathology in Multiple Sclerosis brain” (40,000 Euro)-PI
2010-12 Telethon Grant “Developing new treatments for CMT1X neuropathy” (100,000 Euro)-PI
2011-13 Research Grant of the Research Promotion Foundation: “Gene therapy for CMT1X inherited demyelinating neuropathy” (180,000 Euro)-PI

2011-16 Research Grant, European Leukodystrophy Association (ELA) entitled "Gene therapy for hypomyelinating leukodystrophy" (194,000 Euro for 3 years)- PI

2013-15 Research Grant Award from the Muscular Dystrophy Association, USA (MDA) entitled "Mechanisms of CNS disease in X-linked Charcot-Marie-Tooth Disease" (100,200 US Dollars for Kleopa lab for 2 years)- co-PI

2013-16 Research Grant Award (277250) from the Muscular Dystrophy Association, USA (MDA) entitled "Developing Gene Therapy for Inherited Neuropathy" (280,945 US Dollars for 3 years)- PI

2013-17 Research Grant (RG 3457A2/1) National Multiple Sclerosis Society "Roles of Cx32 and Cx47 in oligodendrocytes" ($ 240,830 USD for Kleopa lab)-co-PI

2016-18 Research Grant from the Charcot-Marie-Tooth Association (CMTA) "Evaluating the outcome of a gene replacement approach in a model of CMT4C neuropathy" ($110,424 for 2 years)- PI

2016-18 Research Grant from the French Muscular Dystrophy Association-AFM - "A gene therapy approach for treating CMT4C neuropathy" (€110,000 for 2 years)-PI

2017-19 Research Grant Award (480030) from the Muscular Dystrophy Association, USA (MDA) – co-funded by the CMT Association- entitled "Expanding the gene therapy approach for treating CMT1X" ($120,000)- PI

2019-22 Research Grant Award (603003) co-funded by the from the Muscular Dystrophy Association, USA (MDA) –CMT Association- entitled "A translatable gene therapy approach to treat CMT1X" ($276,430)- PI

2019-21 Research Grant Award "Development of gene silencing approach to treat CMT1A" – CMT Research Foundation ($99,000, PI).

2019-21 Research Grant from the Charcot-Marie-Tooth Association (CMTA) "AAV mediated Gene Therapy for CMT4C" ($122,100 for 2 years)- PI

2020-22 Sponsored Research Agreement with Gene Therapy Company ($240,000) (PI), for translating gene therapy for CMT1X

2020-23 Funding by Association Piccolo Grande Guerriero Odv Italy- Research Project Gene therapy for HLD2 leukodystrophy (€85,000) – PI

2021-22 Sponsored Research Agreement with Gene Therapy Company ($97,000) (PI)- for translating gene therapy for CMT4C

2021-23 Research Grant, CMTA "Schwann cell-targeted gene therapy approaches to treat CMT1A and other demyelinating neuropathies" ($38,500 for 18 months)- PI

2022-23 Sponsored Research Agreement with Gene Therapy Company ($132,360) (co-PI)- for translating gene therapy for CMT1A

2023-25 Sponsored Research Agreement with Gene Therapy Company ($341,400) (PI): "A dose escalation study using AAVrh10 or AAVrh74 to treat CMT1X

2023-26 Research Grant, MDA/CMTA "Nanoparticle-based gene delivery to Schwann cells for treating CMT disease" ($299,992.00 for 3 years)-co-PI

2024-26 Research Grant European Leukodystrophy Association (ELA) entitled "Accelerating gene therapy for PMLD1"" (172,400 Euro for 2 years)- PI

2025-26 Sponsored Research Agreement with Gene Therapy Company ($240,120) (PI): "A dose escalation study using AAVrh74 to treat CMT1X"

INVITED LECTURES AND PRESENTATIONS (*selected from over 60 invited lectures*)

5/2008 **Gordon Research Conference on Myelin, Invited Speaker**: "CNS demyelination in CMT1X patients and in gap junction mutant mice" Il Ciocco, Italy, May 2008

8/2013 Connexin pathology in chronic MS and EAE. **Plenary speaker, 12th MS Workshop** organized by the **Japanese Society of Neuroimmunology** in Fukuoka, Japan

9/2016 **International Society for Neuroimmunology Annual Meeting**, Invited talk: "Involvement of gap junction channels in neuroinflammation" Jerusalem, Israel.

7/2017 ***Invited Plenary Speaker*, Peripheral Nerve Society Annual Meeting**. "Intrathecal gene delivery of GJB1 in animal models of CMT1X". Sitges, Spain

10/2021 ***Invited Speaker, 2021 World Congress of Neurology,*** *Rome, Italy (virtual):* Developing gene therapies for inherited demyelinating neuropathies.

12/2021 Invited Speaker, Hellenic Academy for Neuroimmunology Meeting, Thessaloniki, "The role of oligodendrocyte gap junctions in Neuroinflammation

3/ 2022 **Invited Speaker**, 10th Translational Research Meeting on Peripheral Neuropathies: "Molecular mechanisms of CMT and emerging gene therapies", Paris, France

3/2022 ***Invited plenary Speaker***, Congress on Gene Therapies and Regenerative Medicine, Bahrain: "Gene therapies for inherited demyelinating neuropathies"

5/2022 ***Invited plenary P.K. Thomas lecture***. 2022 **Peripheral Nerve Society** Annual Meeting. Miami, USA. "Gene therapies for inherited demyelinating neuropathies"

6/2022 ***Invited Plenary Speaker***, Netherlands Society for Cell and Gene Therapy Annual Meeting, Lunteren, Netherlands

7/2022 Invited Speaker, 17th **International Congress on Neuromuscular Diseases** (ICNMD 2022), Brussels, Belgium: "Developing Novel Therapies for Inherited Neuropathies"

3/2023 ***Invited Speaker***, **UK Neuromuscular Translational Research Conference**, London, UK: "Genetic therapies for demyelinating CMT neuropathies"

6/2023 Invited Speaker, 1st European CMT Specialist's Conference, Paris, France. Organized by European CMT Federation. "Mobilizing CMT Patients to promote active participation in advancing research and treatment for their disease"

9/2023 *Invited Speaker*, Global CMT Research Foundation Research Convention, Boston, USA: ""CMT1X: the disease and the pathway to treatment""

9/2023 ***Invited Plenary Speaker***, Global CMT Research Foundation Research Convention, Boston, USA: "Clinical trial participation in order in advance research and treatment for CMT neuropathies" and "CMT1X: the disease and the pathway to treatment"

10/2023 Invited Webinar Presenter, European Reference Network Rare Neuromuscular Disorders (ERN Euro-NMD): "Gene Therapies for CMT neuropathies"

2/2024 Invited Speaker, European Reference Network Rare Neuromuscular Disorders (ERN Euro-NMD) Annual Meeting, Paris: "Gene Therapies for peripheral neuropathies: Present and future"

5/2024 ***Invited Plenary Speaker***, Italian Peripheral Neuropathy Society 14th Annual Meeting, Trieste, Italy: "Current progress in gene therapy for CMT neuropathies"

9/2024 Invited Speaker, Global CMTRF Convention, Boston, USA: "Current opportunities and challenges in CMT Gene Therapies"

3/2025 Invited Speaker, Greek Neurological Society Neuropathies Meeting, Athens: "Current developments in inherited neuropathies"

3/2025 Invited Speaker, ERN Euro-NMD Annual Meeting, Essen: "The landscape of Gene Therapies in Europe"