


Scientific Program of NBIA 2024


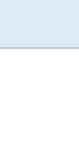
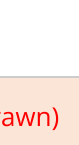


PRESYMPOSIUM COURSE ON NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)

October 17, 2024 (Thursday)

10:00	Registration
12:10	Opening of the Course Bülent Elilbol, Murat Terzi (President of Turkish Neurological Society)
	1. Session: General Perspective of Neurodegeneration with Brain Iron Accumulation (NBIA) Chairs: Bülent Elilbol, Murat Emre
12:20 - 12:50	Introduction of NBIA Zuhail Yapıcı
12:50 - 13:20	Clinical Pearls: Recognizing NBIA Patients Kailash Bhatia (UK)
13:20 - 13:50	Clinical Characteristics and Longterm Follow-up of MPAN Okan Doğu
	2. Session: Medical Treatment and Botulinum Toxin in NBIA Chairs: Bülent Elilbol, Sibel Erten
13:55 - 14:20	Case Discussions Banu Özen Barut, Zeliha Matür, Gençer Genç
14:20 - 14:40	 Coffee Break
	3. Session: Deep Brain Stimulation (DBS) in NBIA Chairs: Çenk Akbostancı, Atilla Yılmaz
14:40 - 15:00	DBS in NBIA from a Neurosurgeon's Perspective Atilla Yılmaz
15:00 - 15:15	Presentation of Turkish Cohort of NBIA with DBS Özgür Öztop Çakmak
	4. Session: DBS Round Table and Case Discussions Chairs: Bülent Elilbol, Ersoy Kocabaşık
15:15 - 16:30	Özgür Öztop Çakmak, Ayşeğül Gündüz, Bilge Koçer, Gül Yalçın Çakmaklı, Pınar Topaloğlu, Zeynep Tufekçioğlu, Esra Erkoç Ataoğlu
	5. Session: Electrophysiological Aspects of NBIA Chairs: Barış Baslo, Neres Bebek
16:30 - 16:50	Peripheral Nervous System Involvement and EMG Findings in NBIA Sadika Özdemir
16:50 - 17:00	Case Presentation: Sezin Baslo
17:00 - 17:20	Epilepsy and EEG in NBIA Ayşe Deniz Elmali
17:20 - 17:40	Highlights and Closing of the NBIA Course
18:30 - 22:00	Welcome Reception for the 9th INTERNATIONAL SYMPOSIUM ON NBIA

9th INTERNATIONAL SYMPOSIUM ON NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)

October 18, 2024 (Friday)

07:00	Registration
08:00	Welcome Address
	Neurodegeneration with Brain Iron Accumulation (NBIA): General Chairs: Murat Emre, Thomas Klopstock
	From Eponyms to Mechanisms: A Century of NBIA Research Guest of Honor: Adrian Danek (Germany)
08:30 - 10:00	Entering the Therapeutics Era for NBIA Disorders Susan Hayflick (USA)
	The TIRCON Registry - Current State and Future Prospects Thomas Klopstock (Germany) Zuhail Yapıcı and Thomas Klopstock
	 Coffee Break
	Oral Sessions Chair: Pervin İşeri
	Group A
	NBIA Diagnosis: Lessons from Ten Year Experience in A French Reference Lab EP1 Valeria Giolosa
	Transcranial Sonography in Neurodegeneration with Brain Iron Accumulation Disorders EP2 Elahe Amini
	Estimation of Ambulation and Survival in Neurodegeneration with Brain Iron Accumulation Disorders EP3 Mohammad Rahani
	Changes in Oxytocin and Oxytocin Receptor Levels in the NBIA Cell Model EP4 Magdalena Zabrska
10:00 - 10:30	Evaluation of Carrier Frequency of Neurodegeneration with Brain Iron Accumulation (NBIA) Disorders in A Middle Eastern Patient Population Based on Data from 17,868 Exome Sequences EP5 Hosan Baş
	Group B
	The Significance of Peroxisomal Dysfunction in the Pathogenesis of NBIA EP6 Karolina Wisniewska
	Survey of NBIA Caregiver Perspectives to Identify Relevant QoI Outcomes for Caregivers/Patients EP7 Janani Ramamurthy
	The Significance of Ferroptosis Dysregulation in The Pathogenesis of NBIA EP8 Karolina Piernynowska
	Initiation and Elongation of the Autophagy Process in Different Types of NBIA EP9 Aneta Szulc
	Is There Any Correlation Between Amount of Iron Measured by Quantitative Susceptibility Mapping and the Clinical Features in NBIA? EP10 Özge Uygun
10:30 - 12:00	PLA2G6-Associated Neurodegeneration (PLAN) Chairs: Murat Emre, Manju Kurian
	The Phenotype and Genotype of PLA2G6-Related Parkinsonism Kailash Bhatia (UK)
	Preparing for Gene Therapy for PLAN Manju Kurian (UK)
	Animal Models for the Investigation of Gene Therapy in INAD Ahad Rahim (UK)
12:00 - 13:00	 Lunch
	Pantothenate Kinase-Associated Neurodegeneration (PKAN) Chairs: Thomas Klopstock, Ody Sibon
	From Fruit Flies to Patients in Less Than 5 Years Ody Sibon (The Netherlands)
13:00 - 14:50	PKAN hiPS-derived Astrocyte Models Highlight the Molecular Mechanism of Iron Accumulation Sonia Levi (Italy)
	Validating Clinical and Ophthalmology Outcome Measures for PKAN Robert W Spaull (UK)
	Clinical Aspects of PKAN and Possible Treatment Based on Deep Brain Stimulation Giovanna Zorzi (Italy)
	Exploring New PKAN Therapies: Current Efforts & Challenges Choukri Ben Mamoun (USA)
	 Coffee Break
	Oral Sessions Chair: Pervin İşeri
	Group A
	Pank Activators for the Treatment of Pantothenate Kinase-Associated Neurodegeneration. (Withdrawn) EP11 Jessica Regan
	Genetic Architecture of Neurodegenerative Disorder and Its Association with Consanguinity in Pashtoon Population EP12 Shoab U Rehman
	The Identification of A 4'-Phosphopantetheine Transporter in S. Cerevisiae EP13 Jouke Jan Wedman
14:50 - 15:20	Investigating Copan in Mice: From Patho-mechanisms to Therapeutic Approaches EP14 Chiara Cavestro
	Group B
	A Surprising Presentation of Atypical Pantothenate Kinase Associated Neurodegeneration Disorder: Metamorphosis EP15 Elahe Amini
	The Effect of Genistein on the Autophagy Process in a Cellular Model of Pantothenate Kinase-Associated Neurodegeneration EP16 Estera Rintz
	Neurodegeneration with Brain Iron Accumulation Disorders and Retinal Neurovascular Structure EP17 Elahe Amini
	Olfactory Status in Neurodegeneration with Brain Iron Accumulation Disorders EP18 Elahe Amini
15:20 - 16:30	Beta-Propeller Protein-Associated Neurodegeneration (BPAN) Chairs: Bülent Elilbol, Sibel Uğur İşeri
	Human Wipi β-Propeller Function in Autophagy and Neurodegeneration Tassula Proikas-Cezanne (Germany)
	Modulating Iron Homeostasis, Autophagy and Oxidative Stress Pathways in BPAN-derived fibroblasts: Active Compounds for Potential Therapy Petek Ballar Kırmızıbayrak (Türkiye)
	Cardiac Glycosides for the Treatment of BPAN and Delineation of the Radiological Phenotype of BPAN Apostolos Papandreou (UK)
	 Coffee Break
	Oral Sessions Chairs: Yıldız Değirmenci, Murat Gültekin
	Group A
	Investigating Respiratory Chain Complex I Deficiency in iPSCs-Derived Dopaminergic Neurons and Astrocytes from BPAN Individuals EP19 Gemma Gasparini
	Characterizing the Molecular Mechanisms of Wipi2 and Wipi4 Mutations in BPAN and Neurodevelopmental Disorders EP20 Eleanor Attridge
16:30 - 17:00	Genistein as an Autophagy Stimulation in Beta-Propeller Protein-Associated Neurodegeneration (BPAN) EP21 Zuzanna Cyske
	Establishing of Drosophila Model for BPAN Disease EP22 Cella Marion
	Group B
	Autophagy and Neurodegeneration: Discovering Critical Insights and Innovative Therapeutic Strategies Using a Novel Human Neuron Model of BPAN EP23 Hetavi Zaveri
	Beta-Propeller Protein Associated Neurodegeneration (BPAN): Electroencephalographic Characterization of a Cohort Of 16 Patients EP24 Alexandra Darling
	Altered Autophagy Involved in Neurodevelopment and Neurodegeneration: Study of A Cohort of Patients with BPAN (Beta-Propeller Protein-Associated Neurodegeneration), New Insights and Phenotypes EP25 Alexandra Darling
17:00 - 17:55	Aceruloplasminemia Chairs: Thomas Klopstock, Sonia Levi
	Protein Replacement and Gene Therapy Approaches in Aceruloplasminemia: Studies in Ceruloplasmin KO Mice Massimo Alessio (Italy)
	A Plasma-Derived Ceruloplasmin from the Optimization of Plasma Fractionation Andrea Caricasole (Italy)
	Recombinant Ceruloplasmin as a Tool to Study the Prevalence of Aceruloplasminemia in GnomAD Nicole Zilotto (Italy)
17:55 - 18:15	Travel and Poster Awards Patricia Wood, Fatemeh Mollet
18:15 - 18:30	End of the Day: Urgent Needs and Next Steps Thomas Klopstock
20:00	 Dinner

9th INTERNATIONAL SYMPOSIUM ON NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)

October 19, 2024 (Saturday)

08:30 - 09:15	Kufor-Rakeb Syndrome (KRS) and Fatty Acid Hydroxylase-Associated Neurodegeneration (FAHN) Chair: Bülent Elilbol
	Cellular Aspects of Iron Accumulation in Kufor-Rakeb Syndrome Patients with Different Type of ATP13A2 Mutations Arzu Karabay (Türkiye)
	Patient Led Research Advances in Fatty Acid Hydroxylase-Associated Neurodegeneration (FAHN/SPG35) Sunita Venkateswaran (Canada)
09:15 - 10:25	Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN)-I Chair: Iwona Kurkowska-Jastrzebska
	Alteration of Autophagy and Lipid Metabolism in MPAN: What We Know and Still Don't Know Arcangela Iuso (Germany)
	Elucidating the Role of C19ORF12 in Triglyceride Homeostasis Rajnish Bharadwaj (USA)
	Putative Involvement of Neuroinflammation in MPAN and Potential for Immunotherapy Erdem Tüzün (Türkiye)
	 Coffee Break
	Oral Sessions Chairs: Yıldız Değirmenci, Murat Gültekin
	Group A
	Modeling C19orf12 Deficiency in Zebrafish (Withdrawn) EP26 Barbara Gnutti
	Early-Onset Dementia in a Case of Late-Onset MPAN EP27 Hatice Yulcel
10:25 - 10:45	Autophagy Disturbance in Fibroblasts Derived from Patients Suffering from Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN), A Type of Neurodegeneration with Brain Iron Accumulation (NBIA) EP28 Grzegorz Wegryzyn
	Mitochondrial Membrane-Protein Associated Neurodegeneration (MPAN) Presenting with Parkinsonism Findings: Case Report EP29 İrem Aydin
	Group B
	Mitochondrial Membrane Protein-Associated Neurodegeneration Presenting with Parkinsonism: A Case Report EP30 Miray Erdem
	Clinical Profile of Kufor-Rakeb Syndrome and Evaluation of Quantitative Sensitivity Analysis EP31 İbrahim Kamacı
	A Case of NBIA with Iron Accumulation Not Detected by Conventional Genetic Testing EP32 Malika Egemberdiyeva
	First Close Look at Proteasome in Neurodegeneration with Brain Iron Accumulation Patients EP33 Lidia Goffke
10:45 - 11:15	Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN)-II Chair: Thomas Klopstock
	MPAN/PKAN Biomarkers Iwona Kurkowska-Jastrzebska (Poland)
	Validated Scale for Neurological Status Assessment in MPAN Patients Marta Skowronska (Poland)
11:15 - 11:40	New Insights from MRI Research in NBIA Chair: Zuhail Yapıcı
	Petr Dušek (Czech Republic)
	COASY Protein-Associated Neurodegeneration (CoPAN) Chair: Valeria Sonia Tiranti, Giovanna Zorzi
11:40 - 12:30	Deciphering COASY-Induced Neurodegeneration: New Variants, New Phenotypes and Insights into the Transcriptomic Profile of Patient Fibroblasts Valeria Sonia Tiranti (Italy)
	COASY-Associated Diseases in Mouse: Not Just a Matter of Neurodegeneration Ivano Di Meo (Italy)
	Zebrafish Models of PKAN and COPAN Daria Finazzi (Italy)
12:30 - 12:45	End of the Day: Urgent Needs and Next Steps Valeria Sonia Tiranti
12:45 - 13:00	Highlights and Closing Remarks