


# Scientific Program of NBIA 2024


## PRESYMPOSIUM COURSE ON NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)



October 17, 2024 (Thursday)

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

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

October 18, 2024 (Friday)

07:00	<b>Registration</b>
08:00	<b>Welcome Address</b> Zuhai Yapıcı and Thomas Klopstock
08:30 - 10:00	<b>Neurodegeneration with Brain Iron Accumulation (NBIA): General</b> <i>Chairs: Murat Emre, Thomas Klopstock</i>
	<b>From Eponyms to Mechanisms: A Century of NBIA Research</b> Guest of Honor: Adrian Danek (Germany)
	<b>Entering the Therapeutics Era for NBIA Disorders</b> Susan Hayflick (USA)
	<b>The TIRCON Registry – Current State and Future Prospects</b> Thomas Klopstock (Germany)
10:00 - 10:30	 <b>Coffee Break</b>
	<b>Poster Sessions</b> <i>Chair: Pervin İşeri</i>
	<b>Group A</b>
	<b>NBIA Diagnosis: Lessons from Ten Year Experience in A French Reference Lab</b> <i>EP1 Valeria Gioiosa</i>
	<b>Transcranial Sonography in Neurodegeneration with Brain Iron Accumulation Disorders</b> <i>EP2 Elahe Amini</i>
	<b>Estimation of Ambulation and Survival in Neurodegeneration with Brain Iron Accumulation Disorders</b> <i>EP3 Mohammad Rohani</i>
	<b>Changes in Oxytocin and Oxytocin Receptor Levels in the NBIA Cell Model</b> <i>EP4 Magdalena Žabiřska</i>
	<b>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</b> <i>EP5 Hasan Bař</i>
	<b>Group B</b>
	<b>The Significance of Peroxisomal Dysfunction in the Pathogenesis of NBIA</b> <i>EP6 Karolina Wiřniewska</i>
<b>Survey of NBIA Caregiver Perspectives to Identify Relevant QoL Outcomes for Caregivers/Patients</b> <i>EP7 Janani Ramamurthy</i>	
<b>The Significance of Ferroptosis Dysregulation in The Pathogenesis of NBIA</b> <i>EP8 Karolina Pierzynowska</i>	
<b>Initiation and Elongation of the Autophagy Process in Different Types of NBIA</b> <i>EP9 Aneta Szulc</i>	
<b>Is There Any Correlation Between Amount of Iron Measured by Quantitative Susceptibility Mapping and the Clinical Features in NBIA?</b> <i>EP10 Özge Uygun</i>	
10:30 - 12:00	<b>PLA2G6-Associated Neurodegeneration (PLAN)</b> <i>Chairs: Murat Emre, Manju Kurian</i>
	<b>The Phenotype and Genotype of PLA2G6-Related Parkinsonism</b> Kailash Bhatia (UK)
	<b>Preparing for Gene Therapy for PLAN</b> Manju Kurian (UK)
	<b>Animal Models for the Investigation of Gene Therapy in INAD</b> Ahad Rahim (UK)

12:00 - 13:00	 <b>Lunch</b>
13:00 - 14:50	<b>Pantothenate Kinase-Associated Neurodegeneration (PKAN)</b> <i>Chairs: Thomas Klopstock, Ody Sibon</i>
	<b>From Fruit Flies to Patients in Less Than 5 Years</b> Ody Sibon (The Netherlands)
	<b>PKAN hiPS-derived Astrocyte Models Highlight the Molecular Mechanism of Iron Accumulation</b> Sonia Levi (Italy)
	<b>Validating Clinical and Ophthalmology Outcome Measures for PKAN</b> Robert VV Spaul (UK)
	<b>Clinical Aspects of PKAN and Possible Treatment Based on Deep Brain Stimulation</b> Giovanna Zorzi (Italy)
<b>Exploring New PKAN Therapies: Current Efforts &amp; Challenges</b> Choukri Ben Mamoun (USA)	
14:50 - 15:20	 <b>Coffee Break</b>  <b>Poster Sessions</b> <i>Chair: Pervin İşeri</i>
	<b>Group A</b>
	<b>Pank Activators for the Treatment of Pantothenate Kinase-Associated Neurodegeneration. (Withdrawn)</b> EP11 Jessica Regan
	<b>Genetic Architecture of Neurodegenerative Disorder and Its Association with Consanguinity in Pashtoon Population</b> EP12 Shoaib Ur Rehman
	<b>The Identification of A 4'-Phosphopantetheine Transporter in <i>S. Cerevisiae</i></b> EP13 Jouke Jan Wedman
	<b>Investigating Copan in Mice: From Patho-mechanisms to Therapeutic Approaches</b> EP14 Chiara Cavestro
	<b>Group B</b>
	<b>A Surprising Presentation of Atypical Pantothenate Kinase Associated Neurodegeneration Disorder: Metamorphopsia</b> EP15 Elahe Amini
<b>The Effect of Genistein on the Autophagy Process in a Cellular Model of Pantothenate Kinase-Associated Neurodegeneration</b> EP16 Estera Rintz	
<b>Neurodegeneration with Brain Iron Accumulation Disorders and Retinal Neurovascular Structure</b> EP17 Elahe Amini	
<b>Olfactory Status in Neurodegeneration with Brain Iron Accumulation Disorders</b> EP18 Elahe Amini	
15:20 - 16:30	<b>Beta-Propeller Protein-Associated Neurodegeneration (BPAN)</b> <i>Chairs: Bülent Elibol, Sibel Uğur İşeri</i>
	<b>Human WIPI <math>\beta</math>-Propeller Function in Autophagy and Neurodegeneration</b> Tassula Proikas-Cezanne (Germany)
	<b>Modulating Iron Homeostasis, Autophagy and Oxidative Stress Pathways in BPAN-derived fibroblasts: Active Compounds for Potential Therapy</b> Petek Ballar Kırmızıbayrak (Türkiye)
	<b>Cardiac Glycosides for the Treatment of BPAN and Delineation of the Radiological Phenotype of BPAN</b> Apostolos Papandreou (UK)



Coffee Break

Poster Sessions

Chairs: Yıldız Değirmenci, Murat Gültekin

Group A

**Investigating Respiratory Chain Complex I Deficiency in iPSC-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

**Genistein as an Autophagy Stimulation in Beta-Propeller Protein-Associated Neurodegeneration (BPAN)**

EP21 Zuzanna Cyske

**Establishing of Drosophila Model for BPAN Disease**

EP22 Celle Marion

16:30 - 17:00

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 Alejandra 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 Alejandra Darling

**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 Ziliotto (Italy)

17:00 - 17:55

**Travel and Poster Awards**

Patricia Wood, Fatemeh Mollet

17:55 - 18:15

**End of the Day: Urgent Needs and Next Steps**

Thomas Klopstock

18:15 - 18:30

20:00



Dinner

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

October 19, 2024 (Saturday)

08:30 - 09:15	<b>Kufor-Rakeb Syndrome (KRS) and Fatty Acid Hydroxylase-Associated Neurodegeneration (FAHN)</b> <i>Chair: Bülent Elibol</i>
	<b>Cellular Aspects of Iron Accumulation in Kufor-Rakeb Syndrome Patients with Different Type of ATP13A2 Mutations</b> Arzu Karabay (Türkiye)
	<b>Patient Led Research Advances in Fatty Acid Hydroxylase-Associated Neurodegeneration (FAHN/SPG35)</b> Sunita Venkateswaran (Canada)
09:15 - 10:25	<b>Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN)-I</b> <i>Chair: Iwona Kurkowska-Jastrzębska</i>
	<b>Alteration of Autophagy and Lipid Metabolism in MPAN: What We Know and Still Don't Know</b> Arcangela Iuso (Germany)
	<b>Elucidating the Role of C19ORF12 in Triglyceride Homeostasis</b> Rajnish Bharadwaj (USA)
	<b>Putative Involvement of Neuroinflammation in MPAN and Potential for Immunotherapy</b> Erdem Tüzün (Türkiye)
10:25 - 10:45	 <b>Coffee Break</b>
	<b>Poster Sessions</b> <i>Chairs: Yıldız Değirmenci, Murat Gültekin</i>
	<b>Group A</b>
	<b>Modeling C19orf12 Deficiency in Zebrafish (Withdrawn)</b> <i>EP26 Barbara Gnutti</i>
	<b>Early-Onset Dementia in a Case of Late-Onset MPAN</b> <i>EP27 Hatice Yuksel</i>
	<b>Autophagy Disturbation in Fibroblasts Derived from Patients Suffering from Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN), A Type of Neurodegeneration with Brain Iron Accumulation (NBIA)</b> <i>EP28 Grzegorz Wegrzyn</i>
	<b>Mitochondrial Membrane-Protein Associated Neurodegeneration (MPAN) Presenting with Parkinsonism Findings: Case Report</b> <i>EP29 Irem Aydin</i>
	<b>Group B</b>
	<b>Mitochondrial Membrane Protein-Associated Neurodegeneration Presenting with Parkinsonism: A Case Report</b> <i>EP30 Miray Erdem</i>
	<b>Clinical Profile of Kufor-Rakeb Syndrome and Evaluation of Quantitative Sensitivity Analysis</b> <i>EP31 Ibrahim Kamaci</i>
<b>A Case of NBIA with Iron Accumulation Not Detected by Conventional Genetic Testing</b> <i>EP32 Malika Egemberdiyeva</i>	
<b>First Close Look at Proteasome in Neurodegeneration with Brain Iron Accumulation Patients</b> <i>EP33 Lidia Gaffke</i>	

	<p><b>Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN)-II</b>  <i>Chair: Thomas Klopstock</i></p>
10:45 - 11:15	<p><b>MPAN/PKAN Biomarkers</b>  Iwona Kurkowska-Jastrzębska (Poland)</p>
	<p><b>Validated Scale for Neurological Status Assessment in MPAN Patients</b>  Marta Skowronska (Poland)</p>
11:15 - 11:40	<p><b>New Insights from MRI Research in NBIA</b>  <i>Chair: Zuhai Yapıcı</i></p>
	<p>Petr Dušek (Czech Republic)</p>
11:40 - 12:30	<p><b>COASY Protein-Associated Neurodegeneration (CoPAN)</b>  <i>Chair: Valeria Sonia Tiranti, Giovanna Zorzi</i></p>
	<p><b>Deciphering COASY-Induced Neurodegeneration: New Variants, New Phenotypes and Insights into the Transcriptomic Profile of Patient Fibroblasts</b>  Valeria Sonia Tiranti (Italy)</p>
	<p><b>COASY-Associated Diseases in Mouse: Not Just a Matter of Neurodegeneration</b>  Ivano Di Meo (Italy)</p>
	<p><b>Zebrafish Models of PKAN and COPAN</b>  Dario Finazzi (Italy)</p>
12:30 - 12:45	<p><b>End of the Day: Urgent Needs and Next Steps</b>  Valeria Sonia Tiranti</p>
12:45 - 13:00	<p><b>Highlights and Closing Remarks</b></p>