

Integrated Neuroscience Session: Mitochondria in Diseases

FRIDAY, APRIL 18 / 1:00 P.M.–5:00 P.M.

Coordinators: Jeffrey M. Vance, MD, PhD, and M. Flint Beal, MD, FAAN

DISCUSSION SESSION / 1:00 P.M.–2:30 P.M. / ROOM 187

- 1:00 P.M.–1:30 P.M.** Primary mtDNA Mutations: 20 Years On *Salvatore Di Mauro, MD, New York, NY*
1:30 P.M.–2:00 P.M. Mitochondria and Neuronal Survival *Discussant TBD*
2:00 P.M.–2:30 P.M. The Role of Mitochondria in Axonal Neuropathies and What Genetics Is Telling Us *Stephan Zuchner, MD, Miami, FL*

POSTER SESSION / 2:30 P.M.–3:30 P.M. / ROOM 187

- IN6-1.001** CoQ10 Deficiency: Clinical and Genetic Studies *Catarina M. Quinzii, Luis Lopez, Ali C. Naini, Salvatore Di Mauro, Michio Hirano*
- IN6-1.002** Deficiency of Cyclophilin D Protects A β -Mediated Mitochondrial and Neuronal Stress in Alzheimer's Mouse Model *Heng Du, Lan Guo, Fang Fang, John Xi Chen, Shidu Yan*
- IN6-1.003** Unusual Clinical and Molecular Presentation of the A8344G Mutation in mtDNA tRNALys (The "MERRF Mutation") *Jiesheng Lu, Sara Shanske, Jorida Coku, Carlos Luciano, Ali Naini, Salvatore Di Mauro, Michio Hirano*
- IN6-1.004** Instability of the Mitochondrial Genome and Neurodegeneration *Federica Invernizzi, Eleonora Lamantea, Giuseppe Lauria, Giuliana Galassi, Marco Onofri, Luigi Palmieri, Antonella Spinazzola, Massimo Zeviani*
- IN6-1.005** WITHDRAWN
- IN6-1.006** Screening for *POLG1* Mutations in a Southern Italian Population *Chiara Criscuolo, Sarah Ammendola, Domenico Cicala, Sandro Banfi, Alessandro Filla*
- IN6-1.007** Slowly Progressive Multisystem Disorder Associated with a G12276A Mutation in the mtDNA tRNA^{Leu} (CUN) Gene *Jorida Coku, Mahsa Mehrazin, Kurenai Tanji, Michio Hirano, Marc Patterson, Ali B. Naini, Sara Shanske, Salvatore DiMauro*
- IN6-1.008** Clinical Features in Five Children with Mutations in ND3, Another mtDNA "Hotspot" *Jorida Coku, Jiesheng Lu, Ali Naini, Sara Shanske, Jong Hee Chae, Kurenai Tanji, Michio Hirano, Salvatore DiMauro*
- IN6-1.009** Phase II Study of the Mitochondrial Antioxidant Mitoquinone for Parkinson's Disease *Barry J. Snow, Fiona L. Rolfe, Michael P. Murphy, Robin A. J. Smith, Michelle M. Lockhart, Christopher M. Frampton, Kenneth M. Taylor*
- IN6-1.010** Clinical and Molecular Features in 27 Patients with Early-Onset Mitochondrial DNA Depletion Syndromes *Antonella Spinazzola, Valeria Massa, Franco Carrara, Rene Santer, Daniela Concolino, Massimo Zeviani*
- IN6-1.011** Characterization of the Ethel Protein by Cellular and Animals Models: Towards an Understanding of Its Role in Ethylmalonic Encephalopathy *Valeria Tiranti, Rossana Mineri, Carlo Viscomi, Cecilia Tiveron, Marco Rimoldi, Massimo Zeviani*
- IN6-1.012** Respiratory Chain Dysfunction and Oxidative Stress Correlate with Severity of Primary CoQ10 Deficiency *Luis Carlos López, Catarina Quinzii Hirano, Jakob Von-Moltke, Ali Naini, Sindu Krishna, Markus Schuelke, Leonardo Salviati, Placido Navas, Salvatore Di Mauro, Michio Hirano*

PLATFORM SESSION / 3:30 P.M.–5:00 P.M. / ROOM 187

- 3:30 P.M.** **IN6-2.001** Mouse Model of Human Mitochondrial DNA Depletion Due to Mitochondrial Thymidine Kinase Deficiency *H. O. Akman, Beatriz Dorado, Luis C. Lopez, Angels Garcia-Cazorla, Saba Tadesse, Eduardo Bonilla, Maya R. Vila, Michio Hirano*
- 3:45 P.M.** **IN6-2.002** Mitochondrial Network Disruption in Cultured Fibroblasts and Myotubes from Patients with a Novel OPA1 Gene Mutation *Marco Spinazzi, Silvia Cazzola, Alessandra Baracca, Giovanna Cenacchi, Luca Scorrano, Bernd Wissinger, Corrado Angelini, Emanuele Loro, Adriana Malena, Lodovica Vergani*
- 4:00 P.M.** **IN6-2.003** Abnormal Mitochondrial Function and Morphology in Fibroblasts of Patients with Early Onset Parkinson's Disease and Two Parkin Mutations *Oliver Bandmann, Stephanie Klaffke, Patrick Sleiman, Nicholas W. Wood, Heather J. Mortiboys*
- 4:15 P.M.** **IN6-2.004** The Presence of Lewy Body Pathology in Parkinsonism with Multiple Mitochondrial DNA Deletions: an Autopsy Case Report *Daita Kaneda, Masayuki Shintaku, Tomonobu Kato, Nobuyuki Oka, Ichizo Nishino, Yu-ichi Goto*
- 4:30 P.M.** **IN6-2.005** Prevention of mtDNA Depletion in Quiescent Skin Fibroblasts from a Patient with a Novel R225W Mutation in the TK2 Gene *Janet L. Oblinger, Cliff Beall, Daniel R. Boué, K. Reed Clark, Zarife Sahenk*
- 4:45 P.M.** **IN6-2.006** OPA1 Mutations Induce Mitochondrial DNA Instability and Optic Atrophy Plus Phenotypes *Valerio Carelli, Pascal Reynier, Maria Lucia Valentino, Pierre Labauge, Christophe Verny, Alain Furby, Guy Lenaers, Rosanna Carroccia, Luisa Iommarini, Chiara La Morgia, Rocco Liguori, Claudia Zanna, Yolanda Campos, Joaquin Arenas, Rafael Garesse, Bernd Wissinger, Dominique Bonneau, Patrizia Amati-Bonneau*