



Poster Catalogue

Moderated Poster Track 1 (Sessions 1-3)

SESSION 1 BASIC SCIENCE, CEREBROVASCULAR DISORDERS, EPIDEMIOLOGY AND FOLLOW-UP, FETAL NEUROLOGY

P01-01 | Clinical features of Neurofibromatosis Type 1 in Slovenian paediatric population

Peter Zabret, Tanja Golli, Damjan Osredkar, **Zvoka Rener-Primec** (SLOVENIA)

P01-02 | The role of Kynurenine pathway metabolites in the mechanism of ketogenic diet therapy

Iwona Zarnowska, Tomasz Zarnowski, Dominika Wrobel-Dudzinska, Tomasz Kocki, Krystyna Mitosek-Szewczyk, Maciej Gasior, Waldemar Anzej Turski (POLAND, USA)

P01-03 | Cerebral Sinovenous Thrombosis in a neonate with late-onset Group B Streptococcus infection

Maria Tsirigotaki, **Pelagia Vorgia**, Nicole-Hilda Anagnostou, Pelagia Plati, Maria Raissaki, Eleftheria Hatzidakis (GREECE)

P01-04 | Isolated Cortical Venous Thrombosis in healthy children; a case report and a literature review

Abdulhafeez M. Khair, Alana E. Salvucci, Badal Jain, Deja Rose, Badal Jain, Stephen Falchek (USA)

P01-05 | Subcortical Band Heterotopia (SBH), a prenatal diagnosis: case report

Aglaë Blauen, Sylvie Degee, An Jansen, Katty Delbecque, Dana Dumitriu, Maria - Roberta Cilio, Marie-Cécile Nassogne (BELGIUM)

P01-06 | Proposal for an epidemiological study in paediatric prolonged disorders of consciousness

Archana Murugan, Anew A. Mallick, Michael Carter, Ingram Wright, Peta M. Sharples (UNITED KINGDOM)

P01-07 | Narcolepsy Type 1 in paediatric patients in Slovenia

Barbara Gnidovec Stražšar (SLOVENIA)

P01-08 | A rare cause of Stroke in childhood: Atrial Myxoma

Didem Ardiçlı, Sibel Akpinar Tekgündüz, Seyma Kayalı, Esra Ülgen Temel, **Deniz Yılmaz** (TURKEY)

P01-09 | Auditory evoked potentials as early detectors of auditory maturation abnormalities in preterm infants with neonatal complications

Dimitra Lekaditi, Anna-Bettina Haidich, Stefanos Sakellaropoulos, Efthimia Vargiami, Oliver Maier, Vasiliios Kimiskidis, Philip Julian Broser (SWITZERLAND, GREECE, HUNGARY)

P01-10 | The safety of antithrombotic therapy in paediatric Cardioembolic Arterial Ischemic Stroke

Elizabeth (Liza) Pulcine, Michael Seed, Leonardo Brando, Manohar Shroff, Sunitha Palasamuam, Mahmoud Slim, Mahenanath Moharir, Gabrielle deVeber, **Nomazulu Dlamini** (CANADA)

P01-11 | A rare cause of acute Vertigo

Filipa Rodrigues, Joana Ribeiro, Andreia Lomba, Joana Castro Marinho, João Sargento-Freitas, Rui Pedro Pais, Conceição Robalo, Mónica Vasconcelos (PORTUGAL)

P01-12 | Paediatric Migraine in the central region of Portugal: a primary health care-based study

Filipe Palavra, Rafaela Lopes, Mariana Mina, Inês Madanelo, Beatriz Silva, Carine Silva, Joana Cebola, Mariana Ribeiro, Lara Tomás, Inês Figueiredo, Luis Amaral, Luis Paixão, Ana Esperança, Vânia Pinto, Inês Rosendo (PORTUGAL)

P01-13 | Moyamoya syndrome in a child with Sickle Cell Anaemia: a cause of the rare event of childhood stroke

Ilias Georgiadis, Eftychia-Maria Kontouri, Anastasia Anastasiou-Katsiardi (GREECE)

P01-14 | Transient Cerebral Arteriopathy in children and adolescents, features of clinical manifestations and course

Inna Shchederkina, Alexander Kessel, Elena Petryaykina (RUSSIAN FEDERATION)

Moderated Poster Track 1 (Sessions 1-3)

P01-15 | Thrombolysis in children and adolescents with Ischemic Stroke. Experience of the Paediatric Stroke Centre of Moscow

Inna Shchederkina, Pavel Svirin, Matvey Livshts, Elena Petryaykina, Andrey Kharkin, Nina Maslova (RUSSIAN FEDERATION)

P01-16 | Prolonged seizures in children-Scottish population based study

Jay Shetty, Clodagh Mitchell, Paul Leonard, Ailidh Ramsay, Libby Chatterton Dickson, Ailsa McLellan (UNITED KINGDOM)

P01-17 | External compression of the internal carotid artery by the hyoid bone – A rare cause of Stroke

Joana Ribeiro, Filipa Rodrigues, João Sargento-Freitas, Joana Pinto, Rui Pais, Isabel Fineza, Mónica Vasconcelos (PORTUGAL)

P01-18 | Clinico-radiological aspects and outcomes in symptomatic and incidentally diagnosed paediatric Moyamoya

Katerina Psychogiou, Maria Lara Buttigieg, Rebecca Keeping, Stavros Stivaros, Ian Kamaly, Dipak Ram, John Grainger, Sofia Douzgou, Amit Herwadkar, Grace Vassallo (UNITED KINGDOM, MALTA)

P01-19 | ABCC6 as an emerging cause of paediatric Stroke: an illustrative case and literature review

Stefano Sartori, Giorgia Martini, Francesco Causin, Laura Baggio, Andrea Pettenazzo, Paolo Simioni, Giorgio Perilongo, Margherita Nosadini, Leonardo Salviati, Irene Toldo (ITALY)

P01-20 | Two children with Cerebral Palsy and Epilepsy due to a mutation in the COL4A1 gene

Lieve Verstraete (BELGIUM)

P01-21 | About some aspects of Hypoxic-Ischemic Encephalopathy among newborns in the Republic of Armenia

Lilit Galstyan (ARMENIA)

P01-22 | Neurological presentations of Egyptian children with Silvery Hair Syndrome: single centre experience

Marwa Abd Elmaksoud, Yasmine El Chazli, Asmaa Elsharkawy, Walaa Shoman, Marwa Eldeeb, Nesrin Gomaa (EGYPT)

P01-23 | Epilepsy and Neurofibromatosis Type 1 in children: epidemiological and clinical study in Croatian population

Matilda Kovac Sizgoric, Goran Krakar, Filip Sabol, Romana Gjergja Juraski, Ljerka Cvitanovic Sojat, Zlatko Sabol (CROATIA)

P01-24 | Quantitative measurement of cerebrovascular reactivity and executive function in children with Moyamoya Disease

Eun Jung Choi, Robyn Westmacott, Mahenanath Moharir, Manohar Shroff, Daune Macgregor, Pradeep Krishnan, Amanda Robertson, Tricia Willia, Gabrielle Deveber, Fenella Kirkham, William Logan, Nomazulu Dlamini (CANADA, UNITED KINGDOM)

P01-25 | Same-day split-bolus Acetazolamide challenge brain perfusion SPECT in paediatric neurovascular practice

Robert Spaull, Robin Holmes, Marcus Bradley, Todd Smallbone, M. Teo, Richard Edwards, A.A Mallick (UNITED KINGDOM)

P01-26 | Focal Cerebral Arteriopathy in persistent Varicella Zoster Virus (VZV) Central Nervous System (CNS) Infection

Sadaf Ahsan, Ala Fadilah, Min Ong, Vijeya Ganeshan, Fiona Shackley (UNITED KINGDOM)

P01-27 | Moyamoya Syndrome as a rare vasculopathy of the Central Nervous System in children with Neurofibromatosis Type 1

Sanja Delin, Matilda Kovač Šižgorić, Goran Krakar, Vlasta Đuranović, Zlatko Sabol (CROATIA)

P01-28 | Post-neonatal Stroke in children: present and future

Silvana Bulearca, Adelina Glangher, Niculina Butoianu, Carmen Burloiu, Diana Barca, Ioana Minciuc, Oana Tarta-Arsene, Carmen Sandu, Cristina Pomeran, Cristina Maier, Mircea Medrea, Catrinel Iliescu (ROMANIA)

Moderated Poster Track 1 (Sessions 1-3)

P01-29 | Predictors of outcome of Arterial Ischemic Stroke in children

Tatia Aprasidze (GEORGIA)

P01-30 | Epidemiological analysis of Febrile Seizure cases during 2018 in a general hospital of Central Macedonia, Greece

Vagelina Bechlivanli, Georgios Katsaras, Evlampia Tsentemidou, Konstantina Verikkou, Anastasia Batsiou, Eleni Lazaridi, Evangelos Oikonomou, Triantafyllia Tsaprouni (GREECE)

P01-31 | Multicystic Encephalomalacia in a twin: case report and review of the literature

Fenella Kirkham, Vineela Mandava, Harriet Joy, Sanjay Salgia (UNITED KINGDOM)

P01-32 | Characteristics, clinical features and prognosis of presumed Perinatal Stroke

Ayse Aksoy, Ozge Kucur, Busranur Cavdarli, Ali Fettah (TURKEY)

P01-33 | A paediatric case of bilateral Internal Carotid Arteries (ICA) Agenesis who presented with multiple cerebral infarctions

Dana Kim, Hyunji Ahn, Mi-Sun Yum, Hyun-Jin Kim, Tae-Sung Ko (REPUBLIC OF KOREA)

P01-34 | Febrile Seizures: Scottish population based study

Libby Chatterton Dickson, Ailsa McLellan, Paul Leonard, Jay Shetty (UNITED KINGDOM)

P01-35 | Paediatric Stroke following Ergotamine: a case report

Nadja Bednarczuk, Ming Lim, Ata Siddiqui, Karine Lascelles (UNITED KINGDOM)

P01-36 | Improving the quality of life for children with Epilepsy and their caregivers in Greece: a meta-analysis of the first paediatric 24/7 epilepsy hotline (CEH) data

Malvina Vamvakari, Artemis Stephanede, Maria Tsirouda, Melpomeni Giorgi, Maria Spanou, Vassiliki Papaevangelou, Argirios Dinopoulos (GREECE)

P01-37 | Central Nervous System Vasculitis in a boy with Epstein–Barr virus-associated T/Natural Killer-Cell Lymphoproliferative Disorders: a case report

Yun Jeong Lee, Su-Kyeung Hwang, Soonhak Kwon, Jiwon Lee (REPUBLIC OF KOREA)

P01-38 | Training programme for rescue treatment of seizures at school environment

Maria Tsirouda, Eleftheria Thoma, Konstantinos Tsoumacas, Maria Spanou, Melpomeni Giorgi, Achilleas Attilakos, Vassiliki Papaevangelou, Argirios Dinopoulos (GREECE)

SESSION 2 EPILEPSY: DIAGNOSIS AND INVESTIGATIONS

P02-01 | Refractory infantile spasms in a boy with Down Syndrome and Koolen-De Vries Syndrome

Katarina Esih, Zvonka Rener-Primec (SLOVENIA)

P02-02 | Co-existence of Type 1 Diabetes Mellitus and Periventricular Heterotopia in a child: a case report

Faruk Inceci, Fatih Gurbuz (TURKEY)

P02-03 | Whole Exome Sequencing (WES) limitations in diagnosis of early Infantile Encephalopathy

Pelagia Vorgia, Maria Tsirigotaki, Eleftheria Papadopoulou, Eleftheria Hatzidakis (GREECE)

Moderated Poster Track 1 (Sessions 1-3)

P02-04 | The clinical difference between infants with PRRT2-Positive and infants with PRRT2-Negative Cluster Epilepsy

Aalt van Roest, Anouk Van Der Vel, Berten Ceulemans (BELGIUM)

P02-05 | Muscarinic Acetylcholine receptor M1 mutation causing early Infantile Epileptic Encephalopathy

Alfons Macaya, Anna Marcé-Grau, Xabier Elorza, Carla Pérez-Rius, Raúl Estévez (SPAIN)

P02-06 | Mutation in the KCNQ2 ion selectivity filter causing severe Epileptic Encephalopathy responsive to Retigabine

Andreea Nissenkorn, Polina Kornilov, Gali Heimer, Bruria Benzeev, Bernard Attali (ISRAEL)

P02-07 | Therapeutic drug monitoring of Fenfluramine and its metabolite Norfenfluramine in patients with Dravet Syndrome

An-Sofie Schoonjans, Laurence Roosens, Berten Ceulemans (BELGIUM)

P02-08 | Reflex Hot Water Epilepsy in children: Spanish multicentre retrospective study

Antonio Hedrera Fernandez, M. Angeles Perez Jimenez, Ramon Cancho Candela, Teresa De Santos Moreno, Juan Jose Garcia Peñas, M. Luz Ruiz-Falco Rojas, Marta Garcia Fernandez, Monica Cano Del Pozo, Raquel Blanco Lago, Ignacio Malaga Dieguez (SPAIN)

P02-09 | ALG13 de novo pathogenic mutation in a male child with Congenital Heart Disease and vertebral abnormalities

Birendra Rai, Muhammad Imran Riazat, Farhana Sharif (UNITED KINGDOM, IRELAND)

P02-10 | Diagnostic rate of targeted gene panel analysis of patients with early-infantile Epileptic Encephalopathy

Bülent Kara, Deniz Sünnetiç Akköynülu, Ayfer Sakarya Güneş, Mesut Güngör, Naci Çine (TURKEY)

P02-11 | Alternative approaches to conventional antiepileptic drugs in early infantile Epileptic Encephalopathies: Galactose and Inositol therapies for SLC35A2 and PLCB1 gene mutations

Bülent Kara, Deniz Sünnetiç Akköynülu, Melis Kavrak, Derya Seven Karaman, Ayfer Sakarya Güneş, Mesut Güngör, Naci Çine (TURKEY)

P02-12 | Neonatal Epilepsy and genetic testing: a new approach

Elena Maqueda Castellote, Montserrat Garcia Puig, Neus Baena, Anna Ruiz, Miriam Guitart, Pablo Ruiz, Carme Figaró, Jana Dominguez (SPAIN)

P02-13 | Utility of gene panel testing in children with seizure onset after 2 years of age: results from a European and Middle Eastern Epilepsy genetic testing programme

Kimberly Gall, **Emanuela Izzo**, Nicole Miller, Kirsi Alakurtti, Eija Seppala, Lotta Koskinen, Juha Koskenvuo, Tero-Pekka Alastalo (USA)

P02-14 | Infantile spasms without Hypsarrhythmia – A distinct Epileptic Syndrome? – A series of 5 patients

Evelina Iachim, Smaranda Nita, Eugenia Roza, Magdalena Sandu, Daniela Vasile, Raluca Teleanu (ROMANIA)

P02-15 | A case at the crossroads of Neurology and Cardiology

Evelina Iachim, Eugenia Roza, Smaranda Nita, Raluca Teleanu (ROMANIA)

P02-16 | Reflex seizures in "Benign Myoclonic Epilepsy in Infancy"

Gorande Kanabar, Inyang Takon, Niju Jacob, Stewart Boyd (UNITED KINGDOM)

Moderated Poster Track 1 (Sessions 1-3)

P02-17 | Chromosome microarray in children with Epilepsy and neurological abnormalities – Our experience
Jelena Radic Nisevic, Ivana Kolic, Melani Mamic, Igor Prpic (CROATIA)

P02-18 | SLC6A1 variant in a child with Intractable Epilepsy

Kristina Lotha, Lydia M.C. Green, Salim Uka, Anthony Hamilton, Martin Tisdall, Gayatri Vadlamani (UNITED KINGDOM)

P02-19 | The risks of Epilepsy after the first attack in children

Leanid Shalkevich, Iryna Zhauniaronak (BELARUS)

P02-20 | Cerebral Palsy and Epilepsy

Madina Taghiyeva (AZERBAIJAN)

P02-21 | Evaluation of the cognitive side effects of antiepileptic treatment in children and adolescents with Epilepsy by the Test Epitrack® Junior

Maria Papadopoulou, Isabella Coco, Faustine Ilski-Lecoanet, Clara Milleret, Joseph Toulouse, Eleni Panagiotakaki, Alexis Arzimanoglou (FRANCE)

P02-22 | Infantile spasms – How often do we think of vitamin B12 deficiency

Monika Kukuruzovic, Masa Malenica, Iva Separovic (CROATIA)

P02-23 | Successful Epilepsy surgery outcome in a child with Mesial Temporal Sclerosis whose initial EEGs were suggestive of benign Focal Epilepsy

Munni Ray, Gayatri Vadlamani, Matthew Morrell, Daniel Warren, Paul Chumas (UNITED KINGDOM)

P02-24 | CLN5 mutation causing Neuronal Ceroid Lipofuscinosis: a case report

Naz Kadem, Gultekin Kutluk (TURKEY)

P02-25 | Clinical and neuroimaging findings in patients with benign Focal Epileptiform Discharges of childhood
Neslihan Bilgin, K. Karli Oguz, Meral Topcu, Dilek Yalnizoglu (TURKEY)

P02-26 | Paediatric Status Epilepticus: a retrospective overview

Özlem Yayıci Köken, Aysegül Danış, Mehmet Fatih Akif Özdemir, Özge Kucur, Ülkühan Öztoprak, Çiğdem Genç Sel, Erhan Aksoy, Ayşe Aksoy, **Deniz Yüksel** (TURKEY)

P02-27 | Fatal Status Epilepticus in Dravet Syndrome: an acute Encephalopathy triggered by fever

Paola De Liso, Virginia Pironi, Massimo Mastrangelo, Domenica Battaglia, Pasquale Striano, Dana Craiu, Federico Vigevano (ITALY, ROMANIA)

P02-28 | Bisphenol A levels in children with acute seizures

Elif Perihan Oncel, Nihal Olgac Dundar, Pinar Gencpinar, **Pinar Arican**, Seher Jabbari Behrouz, Fatma Demet Arslan, Bumin Nuri Dundar (TURKEY)

P02-29 | A new cause of electrical Status Epilepticus in sleep: CDKL5 Disorder

Pinar Arican, Pinar Gencpinar, Nihal Olgac Dundar (TURKEY)

P02-30 | Risk factors of Epilepsy in children with complex febrile seizures; a retrospective cohort study

Chinnuwat Sanguansermsri, **Piyatida Jongruk** (THAILAND)

P02-31 | The aetiology of Epilepsia Partialis Continua: a cautionary tale

Preerna Choudhury, Paula Avram, Johann te Water Naude, Frances Gibbon (UNITED KINGDOM)

Moderated Poster Track 1 (Sessions 1-3)

P02-32 | Jeavons Syndrome: challenges in diagnosis and treatment

Shoaib Khan, Jo Campbell, Ena Cromar, Elma Stephen (UNITED KINGDOM)

P02-33 | Aicardi Syndrome: clinical and neuroradiological phenotype associations

Silvia Masnada, Anna Pichieccio, Mara Cavallin, Manuela Formica, Romina Romaniello, Renato Borgatti, Filippo Arrigoni, Patrizia Accorsi, Lucio Giordano, Lorenzo Pinelli, Martino Montomoli, Renzo Guerrini, Martina Mortilla, Paolo Bonanni, Elisa Osanni, Alberto Danieli, Francesca La Briola, Aglaia Vignoli, Federico Vigevano, Pasquale Parisi, Alessandro Ferretti, N. Deconinck, Francesca Darra, Bernardo Dalla Bernardina, Alexis Arzimanoglou, Valentina De Giorgis, Emilio Perucca, Paola Borrelli, Nadia Bahi-Buisson, Pierangelo Veggiotti, Aicardi Syndrome International Study Group (ITALY, FRANCE, BELGIUM)

P02-34 | A novel DEPDC5 mutation and a known pathogenic mutation in CLCN1 gene in a family with Epilepsy and a variable phenotype

Stella Mouskou, Katherine Anagnostopoulou, Sotiria Mastroyianni, Konstantinos Voudris, George Vartzelis, Anastasia Korona, Maria Mpalla, Harry Kontos, Efstathia Katsarou-Pectacides (GREECE)

P02-35 | Case of Focal Frontal Epilepsy in a 3 months-child with mutation of the NRPL3 gene

Tetiana Stetsenko (UKRAINE)

P02-36 | Electroencephalographic changes in children with Autism Spectrum Disorders

Tetyana Yanina, Olga Rybalko, Eugene Krasnov (RUSSIAN FEDERATION)

P02-37 | Use of SISCOM for the identification of the epileptogenic zone in children

Thomas Foiadelli, Lieven Lagae, Karolien Goffin, Mara De Amici, Lucia Sacchi, Salvatore Savasta, Katrien Jansen (ITALY, BELGIUM)

P02-38 | Psychogenic Non-epileptic Seizures (PNES): a retrospective study on outcomes in a general hospital paediatric service in the United Kingdom

Vivien Wong-Spracklen, Jill Conium, Inyang Takon (UNITED KINGDOM)

P02-39 | Changes in background electroencephalographic activity in benign childhood Epilepsy with centrotemporal spikes after Oxcarbazepine treatment: a Standardized Low-Resolution Brain Electromagnetic Tomography (sLORETA) study

Ye-Hwa Jun, Tae-Hoon Eom, Young-Hoon Kim, Seung-Yun Chung, In-Goo Lee, Jung-Min Kim (REPUBLIC OF KOREA)

P02-40 | DHX30 gene mutations: a new cause of Neurodevelopmental Disorder associated with refractory seizures

Joana Mecca, Luciana Rocha, Zumira Carneiro, Charles Lourenco (BRAZIL)

P02-41 | Expanding the clinical phenotype of DEAF1-Associated Neurodevelopmental Disorder (DAND) gene mutations: two new cases displaying different neurological presentations

Luciana Rocha, Joana Mecca, Laura Vagnini, Zumira Carneiro, Leila Azevedo, Charles Lourenco (BRAZIL)

P02-42 | Problems of sustainability of rendering medical assistance to patients sufficiating Epileption during transition them from the children's network to adult practice

Sergey Burd, Mikhail Mironov, Marina Sarzhina, Marina Gunchenko, Anna Lebedeva, Yuliya Rubleva, Tatyana Krasilshikova, Tatyana Batysheva (RUSSIAN FEDERATION)

P02-43 | Investigating structural brain abnormalities on Neuroimaging in Paediatric Epilepsy

Tony Yu, Jeremy Jones, Ailsa McLellan, Alan Quigley, Jay Shetty (UNITED KINGDOM)

P02-44 | Spinal Muscular Atrophy and Progressive Myoclonic Epilepsy. Report of a Greek paediatric patient

Maria Spanou, Melpomeni Giorgi, Maria Tsirouda, Katherine Anagnostopoulou, Edward Schuchman, Argirios Dinopoulos (GREECE, USA)

Moderated Poster Track 1 (Sessions 1-3)

P02-45 | Heterogeneity in early Infantile Epileptic Encephalopathies other than SCN1A – Description of 21 cases
Melpomeni Giorgi, George Vartzelis, Maria Spanou, Maria Tsirouda, Anastasia Korona, Katerina Anagnostopoulou, Argirios Dinopoulos (GREECE)

P02-46 | Genetic Epileptic Encephalopathies: review and analysis of diagnostic paradigm from a tertiary centre
Rita Martins, **Sofia Quintas**, Pedro Viana, Tiago Santos, Oana Moldovan, Ana Berta Sousa, Patricia Dias, Joana Coelho, Carla Bentes, Ana Rita Peralta, Teresa Moreno, Isabel Amorim, José Pimentel, António Levy (PORTUGAL)

P02-47 | Soccer and seizures – Case report of teenagers with exercise-induced Epilepsy
Tatjana Kovacevic-Preradovic, Nils Holert, Thomas Mayer (GERMANY)

P02-48 | A severe phenotype of early Infantile Epileptic Encephalopathy associated with mutation in CPLX1
Anna Baró-Serrano, Miquel Raspall-Chaure, Anna Marcé-Grau, Monica Vicente-Rasoamalala, Lucia Dougherty-de Miguel, Laura Costa-Comellas, Tania Estrada-Rivera, David Barzallo-Moscoso, Ana Felipe-Rucián, Alfons Macaya-Ruiz (SPAIN)

P02-49 | 17 cases of late onset spasms
Hikaru Kitahara, Shuichi Shimakawa, Motoko Ogino, Miho Fukui (JAPAN)

P02-50 | Neonatal onset Epileptic Encephalopathy, a new CACNA1C-related phenotype
Lucía Dougherty-de Miguel, Miquel Raspall-Chaure, Ana Felipe-Rucián, Anna Baró-Serrano, Laura Costa-Comellas, David Barzallo-Moscoso, Tania Estrada-Rivera, Anna Marcé-Grau, Mónica Vicente-Rasoamalala, Alfons Macaya (SPAIN)

P02-51 | Diagnostic yield of WES following aCGH in the genetic aetiology of early onset Epileptic Encephalopathies
Dilsad Turkdogan, Ayberk Turkyilmaz, Gunes Sager, Gulten Thomas, Olcay Unver (TURKEY)

P02-52 | SLC13A5-related Encephalopathy with Epilepsy in the neonatal period: remarkable improvement on Carbamazepine and Pyridoxal-Phosphate. What worked out so well?
Roberto Santalucia, Catheline Vilain, Julie Soblet, Corienne De Laet, Aline Vuckovic, Alec Aeby (BELGIUM)

SESSION 3 INFLAMMATORY DISEASES OF THE CNS

P03-01 | Anti-MOG antibody associated Central Nervous System Demyelination following Bartonella Meningo-Encephalitis: a case report
Gavin Fatania, Cillian McNamara, Sara Ffrench-Constant, Leena Mewasingh, Sushil Beri, Brynmor Jones, Carolina Kachramanaoglu, **Wajanat Jan** (UNITED KINGDOM)

P03-02 | The diverse spectrum of Encephalopathy in Acute Paediatric Neurology Services: a multi-centre evaluation

Jonathan Gadian, Michael Perry, Cheryl Hemingway, Marios Kaliakatsos, Shan Tang, **Ming Lim**, On behalf of TRAMPOLINE (TRAnsforming Management through PolyOmic assessment of Life-threatening eNCEphalopathy) study group (UNITED KINGDOM)

P03-03 | Hashimoto Encephalitis: a rare cause of Encephalitis associated with classical clinical profile
Mariane Saliba, Nathalie Cajfinger, Marc Trippaerts, Anne-Simone Parent, **Patricia Leroy** (BELGIUM)

P03-04 | MOG-Ab-associated Disease presenting with relapsing Meningoencephalitis with high intracranial pressure in a four-year-old girl

Renata Paolilo, Polyanna Cerqueira, Ciro Matsui-Junior, Patricia Takahashi, Felipe Souza, Renato Albuquerque, Maria Almeida, Paula Kraichete, Frederico Moura (BRAZIL)

Moderated Poster Track 1 (Sessions 1-3)

P03-05 | Challenges in determining the aetiology and treatment of Acute Flaccid Myelitis

Robert Spaull, Todd Smallbone, Emily Morrow, Anu Goenka, Jolanta Bernatoniene, Charles Irish, Marcus Likeman, Peter Muir, Kayal Vijayakumar (UNITED KINGDOM)

P03-06 | Paediatric Catatonia in a young child with anti NMDAR Autoimmune Encephalitis; a case report and brief literature review

Abdulhafeez M. Khair, Gurcharanjeet Kaur, Sreenath Thati Chganganna, Badal Jain, Alana E Salvucci, Tara Pezzuto, Rawad Obeid, Aparna Polavarapu, Josephine Elia, Michael Cellucci (USA)

P03-07 | An atypical presentation of Miller-Fisher Syndrome in an adolescent

Anastasia Korona, Maria Balla, Stella Mouskou, Aikaterini Markante, Andromachi Stamatou, Eirini Eleftheriou, Sotiria Mastroyianni, Efstatia Katsarou-Pectasides, Konstantinos Voudris, George Vartzelis (GREECE)

P03-08 | NMOSD presenting as combined central and peripheral Demyelination with positive MOG antibodies: a case report

Anastasia Korona, Maria Balla, Stella Mouskou, Lydia Kossiva, Alexandra Soldatou, Ioannis Nikas, Sotiria Mastroyianni, Efstatia Katsarou-Pectasides, Konstantinos Voudris, George Vartzelis (GREECE)

P03-09 | Mild Encephalitis with a Reversible Splenial Lesion (MERS) related to Influenza A Infection in a previously healthy toddler

Antonio Hedrera Fernandez, Raquel Blanco Lago, Gonzalo Anes Gonzalez, Luis Santoveña Gonzalez, Sergio Menendez Cuervo, Nelly Alvarez Alvarez, Silvia Martin Ramos, Laura Nonide Mayo, Virginia Ainhoa Oreña Ansonera, Ignacio Malaga Dieguez (SPAIN)

P03-10 | An interesting case of Tuberculosis Pachymeningitis

Beatriz Borba Casella, Mayara Tiemi Ayres Sakuma, Ciro Matsui Junior, José Albino da Paz, Clarissa Bueno, Umbertina Conti Reed (BRAZIL)

P03-11 | Clinical, therapeutical and evolutive features in Guillain-Barré Syndrome

Catalina Bucur, Carmen Sandu, Silviana Bulearca, Adelina Glangher, Alina Costea, Raluca Tudorache, Catrinel Iliescu, Carmen Burliu, Cristina Pomeran, Ioana Minciù, Dana Craiu, Alice Dica, Diana Barca, Oana Tarta, Cristina Motoescu, Niculina Butoianu (ROMANIA)

P03-12 | A 10-year single centre review of Transverse Myelitis-Plus

Celeste Manchoon, Jonathan Gadian, Ming Lim (UNITED KINGDOM)

P03-13 | Intracranial Hypertension and Papilledema, rare complication of Guillain-Barré Syndrome. Case report

Christina Doxaki, **Pelagia Vorgia**, Eleftheria Papadopoulou, Olga Iliaki, Emmanouel Galanakis (GREECE)

P03-14 | Paediatric anti-MOG antibody-positive cortical Encephalitis – A rare presentation

Cillian McNamara, Gavin Fatania, Sara Ffrench-Constant, Leena Mewasingh, Sushil Beri, Brynmor Jones, Carolina Kachramanoglou, **Wajanat Jan** (UNITED KINGDOM)

P03-15 | A paediatric patient with Anti-N-Methyl-D-Aspartate Receptor Encephalitis: excellent outcome

Dilek Cavusoglu, Nihal Olgac Dundar (TURKEY)

P03-16 | Key clues for diagnosis of Giant Axonal Neuropathy: a case report

Dilek Cavusoglu, Muhsin Elmas, Basak Gogus (TURKEY)

P03-17 | Assessment of the clinical features of Migraine in children with signs of Immune Deficiency

Durdona Aljianova, Yokutkhon Madjidova (UZBEKISTAN)

Moderated Poster Track 1 (Sessions 1-3)

P03-18 | First-line treatment with Natalizumab in paediatric Multiple Sclerosis

Elma Stephen (UNITED KINGDOM)

P03-19 | Evolution of the neuroimaging findings in a patient with Enteroviral Infection: limbic involvement

Esra Serdaroglu, Rahsan Gocmen, Goknur Haliloglu, Banu Anlar (TURKEY)

P03-20 | Acute Postinfectious Cerebellar Ataxia due to Adenovirus

Evangelia Bachlava, Chrysoula Michaletou, Niki Nana, Kleio Daskalaki, Aikaterini Giannisi, Elli Maria Petrou, George Stokidis, Panagiota Athanasopoulou, Marina Katsalouli, Virginia Theodorou (GREECE)

P03-21 | Paediatric-Onset Multiple Sclerosis: 8 years of experience in a tertiary centre

Filipe Palavra, Catarina Fernandes, João Durães, Filipa Rodrigues, Joana Afonso Ribeiro, Isabel Fineza, Conceição Robalo, Carmen Costa, Cristina Pereira, Lívia Sousa, Mónica Vasconcelos (PORTUGAL)

P03-22 | Demyelinating Diseases associated with anti-MOG Antibodies – 3 different cases of an expanding spectrum

Filipe Palavra, Mafalda Cascais, Sofia Costa, Sílvia Carvalho, Júlia Vasconcelos (PORTUGAL)

P03-23 | Panuveitis, CNS involvement and skin manifestations in a 12.5 years old boy with Adamantiades- Behçet's Disease

Vasiliki Koute, Georgia Gazeti, Maria Kotoula, Theodora Simopoulou, George Syrigiannopoulos (GREECE)

P03-24 | Anorexia as presenting symptom of NMDA Receptor Encephalitis

Helene Verhelst, Patrick Verloo, Arnaud Vanlander, Rudy Van Coster (BELGIUM)

P03-25 | Anti-N-methyl-D-Aspartate Receptor Encephalitis treated with Cyclophosphamide using the same protocol in children

Hiromi Aoyama, Taku Omata, Yoshimi Watanabe, Kazuo Kodama (JAPAN)

P03-26 | Safety and efficacy of a Neurokinin-1 receptor antagonist in Subacute Sclerosing Panencephalitis: a Phase 2 randomized clinical trial

Ibrahim Oncel, Mesut Sancar, Bahadir Konuskan, Filiz Ariozi, Emine Arman Kandirmaz, Banu Anlar (TURKEY)

P03-27 | Immunomodulatory treatment of MS may cause progression of NMOSD

Ismail Solmaz, Bahadir Konuşkan, Kader Karlıoğuz, Banu Anlar (TURKEY)

P03-28 | Functional limitations in paediatric-onset Multiple Sclerosis

Ismail Solmaz, Bahadir Konuşkan, Görkem Ertuğrul, Songül Aksoy, Banu Anlar (TURKEY)

P03-29 | Acute Disseminated Encephalomyelitis with longitudinally extensive transverse Myelitis: severe presentation of Yellow Fever vaccine-associated Neurotropic Disease

Jose Albino da Paz, Jorio Mota, Tamisa Sampaio, Mayara Sakuma, Rafaela Ferreira, Ciro Matsui, Renata Paolilo (BRAZIL)

P03-30 | Neurological Involvement of Behçet Disease on Brazilian children

Jose Albino da Paz, Mayana Sakuma, Tamisa Sampaio, Jorio Mota, Rafaela Ferreira, Renata Paolilo, Ciro Matsui, Clovis Silva (BRAZIL)

P03-31 | A case of Anti-NMDAR Encephalitis mimicking Leukodystrophy with poor response to immunosuppressive therapy

Jose Albino da Paz, Rafaela Ferreira, Mayara Sakuma, Tamisa Sampaio, Jorio Mota, Ciro Matsui, Renata Paolilo, Umbertina Reed (BRAZIL)

Moderated Poster Track 1 (Sessions 1-3)

P03-32 | Enterovirus D68 associated Transverse Myelitis: the new Polio of the 21st century?

Kristina Lotha, Haroon Afridi, Lydia M.C. Green, Shanawaz Hussain (UNITED KINGDOM)

P03-33 | ADEM – A paediatric case series in a tertiary care hospital

Learta Alili Ademi (NORTH MACEDONIA)

P03-34 | Two cases of paediatric AQP4-antibody positive Neuromyelitis Optica Spectrum Disorder successfully treated with Tocilizumab

Markus Breu, Sarah Glatter, Romana Höftberger, Michael Freilinger, Karl Kircher, Gregor Kasprian, Rainer Seidl, Barbara Kornek (AUSTRIA)

P03-35 | Handwriting in Paediatric Onset Multiple Sclerosis – Tracing strategy and kinematic analysis

Nikola Ivančević, Marija Novičić, Vera Miler-Jerković, Milica Janković, Dejan Stevanović, Blažo Nikolić, Mirjana B. Popović, Jasna Jančić (SERBIA)

P03-36 | Evaluation of mental state and cognitive functions of children with Radiologic Isolated Syndrome

Nilüfer Okumuş, Bülent Kara, Ayfer Sakarya Güneş, Yonca Anık, Ayşen Çoşkun (TURKEY)

P03-37 | Novel neuroimaging findings associated with Autoinflammatory Interferonopathies

Noelle Enright, Seamus MacFarland, Ethna Phelan, David Webb, Mary O'Regan (IRELAND)

P03-38 | Guillain-Barré Syndrome in children at quaternary level care hospital: prevalence, clinical profile and outcomes, a retrospective chart review

Nozibongo Voxelka, Lawrence Mubaiwa, Pamela Rapiti (SOUTH AFRICA)

P03-39 | Acute Flaccid Myelitis outbreak through 2016-2018: multicentre experience from Turkey

Olcay Ünver, Dilşad Türkdoğan, Serhat Güler, Osman Kipoğlu, Mesut Güngör, Cem Paketçi, Kürşat Çarman Bora, Hülya Maraş Genç, Mehpare Özkan, Elif Karatoprak, Betül Kılıç, Gülsen Thomas, Erhan Bayram, Coşkun Yarar, Hatice Sözen Gülhan, Güneş Sağıer, Ayfer Sakarya Güneş, Pınar Koytak, Evrim Karadağ Saygı, Gazanfer Ekici, Sema Saltık, Mine Çalışkan, Bülent Kara, Uluç Yiş, Nur Aydinli (TURKEY)

P03-40 | What differentiates NMOSD and Multiple Sclerosis in paediatric population ≤ 11 years of age

Renata Paolillo, José Albino da Paz, Samira Luisa Apostolos-Pereira, Carolina Rimkus, Dagoberto Callegaro, Douglas Sato (BRAZIL)

P03-41 | Retrospective analysis of 13 paediatric patients with clinically Mild Encephalitis/encephalopathy with a Reversible Splenial Lesion (MERS)

Ritsuo Nishiuchi, Chiho Tokorodani, Junya Ohira, Kosuke Tamefusa, Akane Kanazawa, Mari Miyazawa (JAPAN)

P03-42 | Paediatric presentation of CASPR2 Encephalitis

Sarolta Dobner, Zoltán Liptai (HUNGARY)

P03-43 | Radiologically Isolated Syndrome in children

Seher Sarı, İsmail Solmaz, Banu Anlar, Neslihan Bilgin, İbrahim Öncel (TURKEY)

P03-44 | 12-year-old girl with possible Vogt-Kayanagi-Harada Disease requiring treatment with systemic corticosteroids and Cyclosporine

Ilknur Erol, Şeyda Beşen, Aysel Pelit, Aytül Noyan (TURKEY)

P03-45 | Poster withdrawn

Moderated Poster Track 1 (Sessions 1-3)

P03-46 | Efficacy of a drug cocktail containing vitamins to prevent Acute Encephalopathy with Biphasic Seizures and Late Reduced Diffusion (AESD)

Taku Omata, Yoshimi Watanabe, Kazuo Kodama, Hiromi Aoyama (JAPAN)

P03-47 | A case of Autoimmune Encephalitis with cerebellar damage associated with anti-GAD65 antibodies in a child

Tetiana Stetsenko, Halyna Fedushka (UKRAINE)

P03-48 | Challenges in the treatment and prognosis of Chronic Opsoclonus-Myoclonus-Ataxia Syndrome-OMAS

Vesna Brankovic-Sreckovic, Natasa Cerovac, Milan Borkovic, Vedrana Milic Rasic (SERBIA)

P03-49 | A 17-year-old girl with Anti-NMDA Receptor Encephalitis with Ovarian Teratoma and pure psychiatric symptoms

Lampros Kousoulos, Andre C.R. Barth, Isabelle Nassenstein, Ralf-Bodo Tröbs, Andrea Tannapfel, Heidi Dercks, Kevin Rostasy, Andreas Wegener-Panzer (GERMANY)

P03-50 | High prevalence of minor neurological dysfunction after Acute Encephalitis in childhood

Heidi Pöyhönen, Sirkku Setänen, Ville Peltola, Tuire Lähdesmäki (FINLAND, SWEDEN)

P03-51 | A rare case of paediatric Chronic Granulomatous Herpes Simplex Encephalitis

Eva Ioannidou, Omar Abdel-Mannan, Helen Payne, Thomas Jacques, Kishtji Mankad, Yael Hacohen, Alasdair Bamford, Marios Kaliakatsos, Sanjay Bhate (GREECE, UNITED KINGDOM)

P03-52 | 10-year-old female with Seronegative Autoimmune Encephalitis due to HHV-7 infection

Evangelos Christou, Sotiria Mastroyanni, Theodora Bachou, Maria Kourousi, Theodoros Tsikrikas, Stella Mouskou, Konstantinos Voudris, Eustathia Katsarou, Dimitrios Delis (GREECE)

P03-53 | Acute Flaccid Paralysis in paediatric patients the last 11 years in the island of Crete

Georgios Ntoulias, Pelagia Vorgia, Christina Kamari, Georgios Niotakis, Georgia Vlahaki, Maria Raissaki, Eufrosyni Papadaki, Ioanna Tritou, Emmanouil Galanakis (GREECE)

P03-54 | Treatment-Refactory CNS Cryptococcosis – What failed?

João Nuno Ferreira de Carvalho, Ana Filipa Lopes, José Paulo Monteiro, Maria José Fonseca (PORTUGAL)

P03-55 | Acute Longitudinal Extensive Transverse Myelitis – A rare clinical manifestation of Lyme Neuroborreliosis

Dilara Füsün Icagasioglu, Nihal Soylu Aydin, Ayse Aralaşmak, Ozden Turel, Akın Iscan (TURKEY)

P03-56 | Progressive Encephalomyelitis with Rigidity and Myoclonus (PERM) associated with Glycine receptor antibodies

Marina Mitrogiorgou, Melpomeni Giorgi, Harry Alexopoulos, Maria Spanou, Maria Tsirouda, Argirios Dinopoulos (GREECE)

P03-57 | Fulminant Susac's Syndrome: case report – A rare immunological disease in comparison to other autoimmune Encephalopathies

Sarah Glatter (AUSTRIA)

P03-58 | Acute Encephalopathy in an adolescent associated with Bartonella Henselae Infection

Tanja Loboda, Mirjana Perković Benedik (SLOVENIA)

P03-59 | Autoimmune Encephalitis. When the medical history and the clinical picture are suspicious, the investigation procedure must go further. A case report

Pelagia Vorgia, Ioannis Goniotakis, Ilianna Maniadaki, Eleni Mihailidi, Pinelopi Paspalaki, Emmanouil Galanakis (GREECE)

Moderated Poster Track 1 (Sessions 1-3)

P03-60 | Immunosuppressive therapy in a five-year-old girl with Parry-Romberg Syndrome

Ilknur Erol, Murat Durdu, Şeyda Beşen, Aytül Noyan (TURKEY)

P03-61 | Clinical and laboratory differential diagnostical aspects of Multiple Sclerosis, Disseminated Encephalitis and Encephalitis in children

Elena Skripchenko, Galina Ivanova, **Natalia Skripchenko**, Anna Syrovtseva, Elena Murina, Galina Zeleznikova, Lidia Alekseeva, Evgeniy Karev, Olga Goleva (RUSSIAN FEDERATION)

P03-62 | The relationship between Herpes Viruses and Endothelium Dysfunction in paediatric CNS Demyelinating Diseases

Elena Skripchenko, **Natalia Skripchenko**, Galina Ivanova, Anna Syrovtseva, Elena Murina, Olga Goleva (RUSSIAN FEDERATION)

P03-63 | A rare case of Acute Demyelinating Encephalomyelitis associated with Kawasaki Disease

Sema Saltık, Ceren Bibinoğlu Amirov, Aysel Güzeler, Serhat Güler (TURKEY)

Moderated Poster Track 2 (Sessions 4-7)

SESSION 4 EPILEPSY: MEDICAL AND SURGICAL TREATMENT, NEURO-ONCOLOGY, NEURO-PsYCHIATRY

P04-01 | KCNT1 related severe Early Onset Epilepsy – A promising example of Precision Medicine

Aharon Schif, Karin Weiss, Tova Hershkovitz, Lilach Shemer-Meiri, Sarit Ravid (ISRAEL)

P04-02 | T2w Hypointense MRI Lesions may represent an early marker of the Epileptogenic Zone in infants with Tuberous Sclerosis Complex. A case series

Barbara Benova, Hanna Mai Hulshof, Pavel Krsek, Kees P.J. Braun, Floor E. Jansen, Maarten Lequin, Martin Kyncl (CZECH REPUBLIC, NETHERLANDS)

P04-03 | Fenfluramine HCl oral solution provides long-term, clinically meaningful ($\geq 50\%$) reduction in seizure frequency in Dravet Syndrome: interim analysis of a long-term open-label extension study

Berten Ceulemans, An-Sofie Schoonjans, Kate Riney, Judith Verhoeven, Antonio Gil-Nagel, Marina Nikanorova, Sameer M. Zuberi, Stéphane Avuin, Milka Pringsheim, Thomas Mayer, Francesca Darra, Pasquale Striano, Joseph Sullivan, Kelly G. Knupp, Linda C. Laux, Ian Miller, Dinesh Talwar, Eric Marsh, Michael Lock, Arnold Gammaitoni, Gail M. Farfel, Arun Mistry, Glenn Morrison, Anupam Agarwal, Bradley S. Galer (BELGIUM, AUSTRALIA, NETHERLANDS, SPAIN, DENMARK, UNITED KINGDOM, FRANCE, GERMANY, ITALY, USA)

P04-04 | Suspected Autoimmune Epilepsy in children

Chrysoula Michaleiou, Evangelia Bachlava, Kleio Daskalaki, Aikaterini Giannisi, Panagiota Athanasopoulou, Elli-Maria Petrou, Georgios Stokidis, Niki Nana, Virginia Theodorou (GREECE)

P04-05 | KCNQ2-Related Disorders: clinical spectrum and response to sodium channel blockers

Didac Casas-Alba, Judith Armstrong, Delia Yubero, Carlos Valera, Àngels García-Cazorla, Carmen Fons (SPAIN)

P04-06 | Adjunctive Perampanel and Health-Related Quality of Life (HRQoL) in paediatric patients (aged 4 to <12 years) with Partial-Onset Seizures (POS) or Primary Generalised Tonic-Clonic Seizures (PGTCS) in Study 311

Elena Arce Portillo, Anna Patten, Manoj Malhotra, Leock Y. Ngo (SPAIN, UNITED KINGDOM, USA)

P04-07 | A boy with Febrile Infection-related Epilepsy Syndrome (FIREs) treated with a Human Recombinant IL-1 receptor antagonist, a promising treatment option?

Elke Braat, Lieven Lagae, Nathalie Goemans, Liesbeth De Waele, Gunnar Buyse, Carine Wouters, Lien De Somer, Katrien Jansen (BELGIUM)

P04-08 | Preliminary results of the project: "Supporting a Pilot Program for the Creation of Medical Centre Specialized in Diagnosis and Therapy for Children with Fetal Alcohol Spectrum Disorder (FASD)"

Elżbieta Stawicka, Anetta Jeziorek, Krystyna Szymanska, Agnieszka Maryniak, Krzysztof Szczęsny, Eliza Kiepura, Magdalena Bednarczyk, Ewa Matachowska, Marzena Król, Karol Scipio del Campo, Artur Bartochowski, Anna Bujwid (POLAND)

P04-09 | Clinical characteristics of Tic Disorders in children

Young Hoon Kim, Eu Gene Park (REPUBLIC OF KOREA)

P04-10 | Long-term efficacy and safety of Eslicarbazepine Acetate in children: an open-label extension following the double-blind, randomized, placebo-controlled study

Fenella Kirkham, Joana Moreira, Luís M. Magalhães, Fábio Ikedo, Helena Gama (UNITED KINGDOM, PORTUGAL)

P04-11 | Neurological and neuropsychological assessment in young children with Isolated Basal Ganglia Stroke

Giulia Prato, Marta Bertamino, Thea Giacomini, Sara Uccella, Sara Signa, Giulia Amico, Domenico Tortora, Ronchetti Anna, Laura Banov, Isabella Ceccherini, Paolo Moretti, Zanetti Alice, Maja Di Rocco, Maria Savina Severino (ITALY)

Moderated Poster Track 2 (Sessions 4-7)

P04-12 | Ketogenic diet experience of our clinic in Epileptic Encephalopathy patients

Gulten Ozturk Thomas, Birsen Demirel, Dilsad Turkdogan, Gunes Sager, Olcay Unver, Gulcan Akyuz (TURKEY)

P04-13 | Specific personality and amygdala volume in Gorlin Syndrome

Hideki Uchikawa, Katsunori Fujii, Tadashi Shiohama, Naoki Shimojo (JAPAN)

P04-14 | Our experience of Perampanel, a newer Antiepileptic Drug (AED), in the treatment of children with difficult to manage Epilepsy – A 4-year retrospective, single quaternary centre study

Jamie Shah, Rajat Gupta (UNITED KINGDOM)

P04-15 | The association of EEG abnormalities and core symptoms in children with Attention-Deficit/Hyperactivity Disorder

Jung-Chieh Du, Ting-Fang Chiu, Kun-Mei Lee, Min Lee (TAIWAN)

P04-16 | Childhood Absence Epilepsy. Does it really have a good prognosis?

Katalin Hollódy, Mónika Kovács, Eszter Nagy (HUNGARY)

P04-17 | Amantadine – An optional treatment for Severe Secondary Myoclonic Epilepsy and Hyperkinetic Dyskinesia

Keren Politi, Hadasa Stern Goldberg, Tohar Rigler (ISRAEL)

P04-18 | Effects of Adjunctive Perampanel on neuropsychological and growth development in paediatric patients (aged 4 to <12 years) with Partial-Onset Seizures (POS) or Primary Generalised Tonic-Clonic Seizures (PGTCS)

Kimford Meador, Anna Patten, Manoj Malhotra, Leock Y. Ngo (USA, UNITED KINGDOM)

P04-19 | Co-infection Neuroborreliosis and Ehrlichiosis mimicking Paraneoplastic Syndrome – Progressive Encephalomyelitis with Rigidity and Myoclonus (PERM) in teenage girl

Lelde Liepina, Jurgis Strautmanis, Guntis Rozentals, Marta Celmina, Anna Grinfelde, Marija Cehovica (LATVIA)

P04-20 | Anti-epileptic drugs do not resolve impaired memory consolidation in Idiopathic Epilepsy Syndromes of childhood

Mailys Rupin, Julie Remaud, Marie Le Roux, Clémence Coiffard, Patrick Van Bogaert (FRANCE)

P04-21 | Potassium Bromide in Dravet Syndrome and Lennox-Gastaut Syndrome: an underutilized option?

Marco Perulli, Ilaria Contaldo, Maria Luigia Gambardella, Michela Quintiliani, Simona Lucibello, Elisa Musto, Charlotte Dravet, Domenica Immacolata Battaglia (ITALY)

P04-22 | «Masks» of Ischemic Stroke in children with malignancies

Natalia Natrusova, Inna Schederkina, Evgeniya Seliverstova, Olga Tiganova, Elena Petriaikina, Konstantin Kondratchik (RUSSIAN FEDERATION)

P04-23 | Use of a medium chain Triglyceride-based food for special medical purposes in children with Epilepsy: compliance, tolerability and acceptability

Natasha Schoeler, Michael Orford, Umesh Vivekananda, Zoe Simpson, Bahee Van de Bor, Hannah Smith, Tricia Rutherford, Maura O'Donnell, Erika Brennan, James McKenna, Bridget Lambert, Tom Barker, Simona Balestrini, Sanjay M. Sisodiya, Simon J.R. Heales, Matthew C. Walker, Helen Cross (UNITED KINGDOM)

P04-24 | Comparison of neurocognitive outcomes between Levetiracetam and Phenobarbital monotherapy for the treatment of neonatal seizures

Pinar Arican, Nihal Olgac Dundar, Neslihan Mete Atasever, Mine Akkaya Inal, Pinar Gencpinar, Dilek Cavusoglu, Sinem Akbay, Hasan Tekgul (TURKEY)

Moderated Poster Track 2 (Sessions 4-7)

P04-25 | Long-term therapeutic effect of Eslicarbazepine Acetate in children: an open-label extension of cognition study in children 6-16 years

Pierangelo Veggiotti, Joana Moreira, Ana Pereira, Fábio Ikeda, Helena Gama, **Fenella Kirkham** (ITALY, PORTUGAL, UNITED KINGDOM)

P04-26 | Study 311: evaluation of adjunctive Perampanel on mental health in children (aged 4 to <12 years) with Partial-Onset Seizures (POS) or Primary Generalised Tonic-Clonic Seizures (PGTCS)

Rohit Shankar, Jay Salpekar, Anna Patten, **Manoj Malhotra**, Leock Y. Ngo (UNITED KINGDOM, USA)

P04-27 | Natural History of "Congenital" Neurofibromatosis Type 2 (NF2)

Stefano Catanzaro, Andrea Praticò, Agata Polizzi, Piero Pavone, Raffaele Falsaperla, Giuseppe Belfiore, Stefano Palmucci, Massimo Di Pietro, Annalisa Chiarenza, Alessandra Romano, Francesco Di Raimondo, Giuseppe Barbagallo, Roberto Altieri, Graziella Poli, Luigi Maiolino, Chiara Amato, Giuseppe Micali, Francesco Lacarrubba, Milena La Spina, Barbara Amato, Chiara Maria Battaglini, Maria Teresa Garozzo, Giuseppe Zampino, Laura Papi, Roberta Sestini, Alessia Gennaro, Santina Cristina Gorgone, Teresa Mattina, Pietro Milone, Martino Ruggieri (ITALY)

P04-28 | Seizures in children with Acute Lymphoblastic Leukaemia: incidence, risk factors, aetiology and prognosis

Stavroula Anastasopoulou, Mats Heyman, Mats Anders Eriksson, Riitta Niinimäki, Sirje Mikkel, Goda E. Vaitkeviciene, Inga Maria Johannsdottir, Ida Hed Myrberg, Olafur Gisli Jonsson, Bodil Als-Nielsen, Kjeld Schmiegelow, Joanna Banerjee, Susanna Ranta, Arja Harila-Saari (SWEDEN, FINLAND, ESTONIA, LITHUANIA, NORWAY, ICELAND, DENMARK)

P04-29 | A retrospective study of effectiveness and compliance of Ketogenic diet in a single unit

Maria Spanou, Sofia Zouganeli, Melpomeni Giorgi, Maria Tsirouda, Kalliopi Kappou, Katerina Kourtesi, Argirios Dinopoulos (GREECE)

P04-30 | Impact of Methylphenidate on cognitive profile in children with Attention Deficit Hyperactivity Disorder

Sophia Bakhtadze, Nana Geladze, Nana Khachapuridze (GEORGIA)

P04-31 | Description of the use of Levetiracetam in a cohort of Neonatal Onset Epileptic Encephalopathies

Marina Caner Faig, Ming Lim, Karin Lascelles, Rahul Singh (UNITED KINGDOM, SPAIN)

P04-32 | Treatment with Adrenocorticotropin hormone in Drug Resistant Epilepsy – Efficacy in very young infants and older children

Dorota Dunin-Wasowicz, Dorota Domanska-Pakiela, Dariusz Chmielewski, Katarzyna Tomaszek (POLAND)

P04-33 | Estimation of Malondialdehyde, total antioxidant capacity, and Selenium levels in serum of intractable epileptic children receiving treatment with Ketogenic diet

Omnia El Rashidy, Mai Yousef, Yassmin El Gendy, Manal Mohsen, Safaa Morsy, Sarah Dawha (EGYPT)

P04-34 | Hemimegalencephaly and Epilepsy

Vlasta Duranovic, Sanja Pejic Rosko, Zrinka Eres, Katarina Vulin, Ana Tripalo Batos, Tonci Grmoja, Ivana Dakovic, Jadranka Sekelj-Fures, Lana Loncar (CROATIA)

P04-35 | A case of rapidly progressing Pineal Cyst – Is it Pineoblastoma?

Rok Kučan, Tadeja Hostnik, Barbara Faganel Kotnik, Mirjana Perković Benedik, Damjan Osredkar (SLOVENIA)

Moderated Poster Track 2 (Sessions 4-7)

SESSION 5 GENETICS

P05-01 | Poster withdrawn

P05-02 | Rare combination of Oculo Cutaneous Albinism and MEGDEL Syndrome in one patient
Ayşe Aysima Özçelik, Kadri Karaer (TURKEY)

P05-03 | Different clinical manifestations of *TREX1* mutation: a case series

Faruk Incecik, Sibel Balci, Rabia Miray Kisla Ekinci, Ozlem M. Herguner, Atil Bisgin, Mustafa Yilmaz (TURKEY)

P05-04 | Mitochondrial DNA Depletion Syndrome 7 (OMIM #271245) – A case report of a Rare Hepatocerebral Disease

Alexandra Wagner, Christian Staufner, Matias Wagner, Wolfgang Rascher, Patrick Morhart, Hanna Müller, Regina Trollmann (GERMANY)

P05-05 | Rare monogenic causes of Microcephaly: clinical and genetic heterogeneity in a Hungarian cohort

Aliz Zimmermann, Tibor Kalmár, Katalin Szakson, Zoltán Maróti, Melinda Zombor, Adrienn Máté, Marianne Berényi, Borbála Telcs, Kinga Hadzsiev, Béla Melegh, László Sztriha (HUNGARY)

P05-06 | TRAPPC12 related progressive Encephalopathy: 2 cases and expanding phenotype

Ayca Dilrubu Aslanger, Emine Demiral, Seyma Sonmez-Sahin, Serhat Guler, Akin Iscan, Sema Saltik, Gozde Yesil (TURKEY)

P05-07 | Cerebellar Atrophy and Cerebellar Cortex Hyperintensity – Think about Christianson Syndrome

Biayna Sukhdyan, Ani Gevorgyan, Elena Okuneva, Eugen Boltshauser (ARMENIA, RUSSIAN FEDERATION, SWITZERLAND)

P05-08 | Poretti Boltshauser Syndrome: a novel variant in *LAMA1* gene

Pinar Edem, Fatma Ceren Sarioglu, Cem Paketci, Erhan Bayram, Ayse Semra Hiz, Uluc Yis (TURKEY)

P05-09 | A novel *CACNA1A* variant in a child with Hemiplegia and Coma – A case report highlighting the importance of early history

Vanita Shukla, Charoula Siafaka, Marcus Likeman, Andrew A. Mallick (UNITED KINGDOM)

P05-10 | Likely pathogenic *GARS* variant associated with infantile onset Spinobulbar Muscular Atrophy

Carol M. Stephens, Andrew Green, John McHugh, Muireann NiChronin, David Webb, A. Dallosso, N. Forrester, Niamh McSweeney (IRELAND, UNITED KINGDOM)

P05-11 | Partial trisomy of chromosome 13 – A rare cause of developmental delay, Epilepsy and brain malformation
Cristina Anghelescu, Sorina Mihaela Papuc, Aurora Arghir, Magdalena Budisteanu (ROMANIA)

P05-12 | Novel mutations in *MCT8* associated with a less severe phenotype of *MCT8* Deficiency

Silvia Masnada, Stefan Groenweg, Veronica Saletti, Luisa Chiapparini, Barbara Castellotti, Ettore Salsano, W. Edward Visser, Davide Tonducci (ITALY, NETHERLANDS)

P05-13 | Pyruvate Carboxylase Deficiency Type A and Type C: a genotype-phenotype correlation

Emanuele Coci (GERMANY)

P05-14 | Hypokalemic Periodic Paralysis due to a new mutation in *CACNA1S* gene

Filipa Rodrigues, Telma Marques, Sónia Regina Silva (PORTUGAL)

Moderated Poster Track 2 (Sessions 4-7)

P05-15 | The Italian cohort of STXBP1 mutated patients: phenotypic spectrum and novel mutations

Ganna Balagura, Francesca Marchese, Federico Zara, Pasquale Striano, LICE collaborative group on STXBP (ITALY)

P05-16 | Jordan's Syndrome: A rare cause of Macrocephaly, Hypotonia and Epilepsy associated with mutation in the PPP2R5D gene

Gautam Ambegaonkar, Elizabeth Radford (UNITED KINGDOM)

P05-17 | Description of the clinical case of Cockayne Syndrome

Marzhan Lepessova, Aya Jalairova, Zhuldyz Nukebayeva, Gaziza Koregen (KAZAKHSTAN)

P05-18 | The phenotype spectrum of PURA Syndrome: report of three cases

Jessica Gencarelli, Davide Colavito, Stefania Bigoni, Elisa Ballardini, Giampaolo Garani, Claudia Ruivenkamp, Raffaella Faggioli, Alberta Leon, Agnese Suppiej (ITALY, NETHERLANDS)

P05-19 | Ataxia as the presenting feature of Juvenile-Onset Alexander Disease

Jin Sook Lee, Jong-Hee Chae (REPUBLIC OF KOREA)

P05-20 | MECP2 gene anomalies are not synonymous with Rett Syndrome

Joana Martins, Ruben Rocha, Cristina Garrido, Inês Carrilho, Sónia Figueiroa, Manuela Santos, Teresa Temudo (PORTUGAL)

P05-21 | Survey concerning ultra-rare diseases and parental understanding

Karen Jahnke, Erik Eklund (SWEDEN)

P05-22 | The puzzling variability of ATP1A3-Related Disease: 5 new mutations resulting in 5 separate phenotypes

Katerina Vezyrogloou, Manju Kurian, Lucinda Carr, Prab Prabhakar, Cheryl Hemingway, Robert Robinson, Christin Eltze, Helen Cross (UNITED KINGDOM)

P05-23 | Genetic analysis of Early-Onset Epileptic Encephalopathies; 8-year experience

Katherine Anagnostopoulou, Argyrios Dinopoulos, Roser Pons, Geogios Vartzelis, Eleni Skouteli, Artemis Gika, Stella Mouskou, Efstatia Katsarou-Pectacades, Eleni Pantazi, Maria Spanou, Melpomeni Georgi, Artemis Stefanidi, Georgios Niotakis, Iliada Nakou, Joseph Kaleyias, Stamatina Poula, Eleftheria Kokkinou, Vasiliki Zouvelou, Harry Kontos (GREECE, UNITED KINGDOM)

P05-24 | A case of Early-Infantile Onset, Rapidly Progressive Leukoencephalopathy with calcifications and cysts caused by mutations in SNORD11B

Kazuo Kodama, Taku Omata, Yoshimi Watanabe, Hiromi Aoyama, Jun-Ichi Takanashi, Kazuhiro Iwama, Takeshi Mizuguchi, Naomichi Matsumoto (JAPAN)

P05-25 | Novel Heterozygous Missense Mutation c.92A>G in the LMNA gene: a case study

Kyung Eun Nam, Dongwoo Lee, Woori Jang, Myungshin Kim, Joo Hyun Park (REPUBLIC OF KOREA)

P05-26 | A first report of homozygous missense mutation in the ADCY5 gene, related to an Autosomal Dominant Dyskinesia Syndrome

Lilach Shemer-Meiri, Amir Peleg, Lena Sagi-Dain (ISRAEL)

P05-27 | Genetic anomalies in siblings with intellectual disabilities

Magdalena Budisteanu, Sorina Mihaela Papuc, Raluca Grozavescu, Carmen Burloiu, Florina Rad, Ioana Minciu, Diana Barca, Andreea Tutulsn-Cunita, Bogdan Budisteanu, Catrinel Iliescu, Iuliana Dobrescu, Ina Focsa, Dana Craiu, Aurora Arghir (ROMANIA)

Moderated Poster Track 2 (Sessions 4-7)

P05-28 | Probable dysfunction of GABA-A receptors in patients with GNAO1-Related Syndromes – An example of four clinical cases

Malgorzata Pawłowicz, Krystyna Szymanska, Hanna Mierzewska, Małgorzata Rydzanicz, Piotr Stawinski, Marta Zawadzka, Maria Mazurkiewicz-Beldzinska, Rafał Płoski (POLAND)

P05-29 | Can the clinical spectrum of Epileptic Encephalopathy associated with SYNGAP1 mutation be extended to include some lysosomal features? – An example of two clinical cases

Malgorzata Pawłowicz, Małgorzata Rydzanicz, Piotr Stawinski, Rafał Płoski (POLAND)

P05-30 | SCN2A mutation: different genotype and phenotype, clinical cases

Nataliya Smulskaya, Iryna Nicoldaenko (UKRAINE)

P05-31 | Expanding the phenotype of PIGA variants in Early Onset Epilepsy

Noelle Enright, Katie Ryan, Jane Cryan, Mary O'Regan (IRELAND)

P05-32 | Late-infantile Neuronal Ceroid Lipofuscinosis presenting in a child of West African ethnicity with a novel TPP1/CLN2 mutation

Robert Spaull, May-Lin Lui, Clare Beesley, Nicholas Kane, Andrew Mallick (UNITED KINGDOM)

P05-33 | Periventricular Nodular Heterotopia with genetic mutation in Filamin A; a rare case

Samyami Chowdhury, Kristian Aquilina, Susan Holder, Ather Ahmed (UNITED KINGDOM)

P05-34 | Clinical Heterogeneity of CACNA1A mutations in childhood including Hyperekplexia and Global Developmental Delay

Saraswathy Sabanathan, Deepa Krishnakumar, Anna Maw, Bina Mukhtyar, Sarju G. Mehta, **Manali Chitre** (UNITED KINGDOM)

P05-35 | RANBP2 mutation in a Turkish child with Recurrent Acute Necrotizing Encephalopathy

Serhat Guler, Gozde Yesil, Cengiz Yalcinkaya, Ayca Aslanger, Ceren Bibinoglu Amirov, Aysel Guzelser, Sema Saltik (TURKEY)

P05-36 | Brain morphologic study in Rett and Rett-Like Syndrome with MECP2 mutation

Tadashi Shiohama, Jacob Levman, Emi Takahashi (JAPAN, USA)

P05-37 | Novel SLC9A6 mutation in a family with X-linked Intellectual Disability, mimicking Angelman Syndrome

Tinatin Tkemaladze, Nana Khachapuriidze, Sophia Bakhtadze (GEORGIA)

P05-38 | Genomics at the paediatric coalface: equipping Australian non-genetic specialist paediatricians with skills to practise in the genomic era

Zoe McCallum, Amy Nisselle, Belinda McLaren, Lisette Curnow, Yana Smagarinsky, Helen Savoia, David Amor, Sue White (AUSTRALIA)

P05-39 | Practical implications of genetic diagnosis through Array-CGH

Alfonso Amado, Ana Ocampo, Diego Sandin, Antia Fiel, Fernanda Taboas (SPAIN)

P05-40 | Whole-exome sequencing as an early diagnostic tool in children with Progressive Neurological Disorders

Juho Aaltio, Anna Kuukasjärvi, Tuula Lönnqvist, Anu Suomalainen, Pirjo Isohanni (FINLAND)

P05-41 | Epileptic-Dyskinetic Encephalopathy caused by a mutation in the SCN8A gene: a case of neonatal onset

Almudena Chacón Pascual, María Concepción Miranda, María Medina, Paula Carrascosa, Estibaliz Barredo, Alba Ramajo, María Vázquez, Yolanda Ruiz, Pedro de Castro (SPAIN)

Moderated Poster Track 2 (Sessions 4-7)

P05-42 | Intellectual disability associated with dysmorphism, cleft palate, congenital heart defect and behavioural anomalies due to a de novo MEIS2 mutation

Andrea Gangfuß, Goekhan Yigit, Janine Altmüller, Peter Nürnberg, Christina Czeschik, Nina Bögershausen, Peter Burfeind, Dagmar Wieczorek, Bernd Wollnik, Ulrike Schara, Alma Küchler (GERMANY)

P05-43 | A case of Eosinophilic Meningitis accompanied by Phaeohyphomycosis due to Exophiala Dermatitidis

Hyunji Ahn, Mi-Sun Yum, Hyun-Jin Kim, Tae-Sung Ko (REPUBLIC OF KOREA)

P05-44 | IRF2BPL mutation causing a neurodegenerative phenotype with Movement Disorder and Bulbar Palsy

Maria Spanou, Katherine Anagnostopoulou, Maria Tsirouda, Meltem Giorgi, Eleni Pantazi, Stamatina Poula, Haris Kontos, Georgios Vartzelis, Argirios Dinopoulos (GREECE)

P05-45 | A Williams-Beuren Syndrome with chromosome 16p12.2-p11.2 deletion or undeclared new syndrome

Olga Rybalko, Nikolay Kaladze, Galina Dosikova, Aleksan Kogutnitskii (RUSSIAN FEDERATION)

P05-46 | A new compound heterozygous mutation in Ataxia-Telangiectasia: a case report

Sema Bozkaya Yılmaz, Nihal Olgac Dundar, Pinar Arican, Atilla Ersen, Pinar Gencpinar (TURKEY)

P05-47 | Rapid onset Coreodystonia without Parkinsonism due to mutation in the ATP1A3 gene

Maria Victoria Tóffoli, Delia Yubero, Verónica Delgadillo, Monserrat Ortega Rodríguez, Juan Darío Ortigoza-Escobar (SPAIN)

P05-48 | PTEN-related Disorders. Phenotypic description in a series of paediatric patients

Maria Victoria Tóffoli, Jaume Campistol, Hector Salvador Hernández, Antonio Martínez Monseny, Carme Fons Estupiñà (SPAIN)

P05-49 | CACNA1A mutation presenting as early onset Developmental Encephalopathy without Epilepsy: presentation of two cases

Alec Aeby, Marvyn Pichueque, Nicolas Deconinck, Julie Soblet, Catheline Vilain (BELGIUM)

P05-50 | New mutation CACNA1A and its clinical manifestations

Biserka Rešić, Edina Karabeg, Jasmina Rešić-Karara, Enes Karabeg (CROATIA, BOSNIA AND HERZEGOVINA)

P05-51 | Two siblings with a novel mutation in RIN2 gene associated with Epilepsy, further expanding the clinical spectrum

Ilknur Erol, Şeyda Beşen, Asc. Özgür Kütük, Özlem Alkan (TURKEY)

P05-52 | A very early-onset Isolated Dystonia associated with DYT1 gene

Ilknur Erol, Şeyda Beşen (TURKEY)

SESSION 6 METABOLIC DISORDERS

P06-01 | Expanding the neurological spectrum of Seipin Deficiency (BSCL2), a complex lipid defect

Alejandra Darling, Mar O'Callaghan, Andrés Nascimento, Carlos Ortez, Daniel Natera, Verónica González, Julieta González, Verónica Delgadillo, Daniela Revilla, Judith Armstrong, Alfonso de Oyarzábal Sanz, David Araujo Vilar, Cristina Jou, Cecilia Jiménez Mallebrera, Angels García-Cazorla (SPAIN)

P06-02 | Sanfilippo Syndrome Type B: a continuum spectrum of clinical phenotypes

Allan Depizol, Ana Paula Bonatto, Lais Carvalho, Zumira Carneiro, Laura Vagnini, **Charles Lourenco** (BRAZIL)

Moderated Poster Track 2 (Sessions 4-7)

P06-03 | Long-term effectiveness of Enzyme Replacement Therapy (ERT) in paediatric patients with Mucopolysaccharidosis Type II

Monica Troncoso, Fernanda Balut, Daniela Munoz, Veronica Saez, Isadora Ruiz, Diane Vergara, Paola Santander, Alejandra Hernandez, Carla Rojas, Scarlet Witting (CHILE)

P06-04 | Stuttering as a prominent clinical feature in an adolescent patient with CLN3 Disease

Sotiria Mastroianni, **Stella Mouskou**, Smaragda Kamakari, Vasiliki Chouliara, Anastasia Korona, Georgios Vartzelis, Efstatia Katsarou, Konstantinos Voudris (GREECE)

P06-05 | AXO-AAV-GM2 for the treatment of GM2 Gangliosidoses: programme overview

Terence R. Flotte, Gavin Corcoran, Paul Korner, J. Fraser Wright, Yi Mo, Jamie Benoit, Heather Gray-Edwards, Douglas R. Martin, Miguel Sena-Esteves (USA)

P06-06 | Asparagine Synthetase Deficiency – A rare neurometabolic disorder with Congenital Microcephaly, early speech delay and Drug Refractory Epilepsy – Experience from Oman

Amna Al Futaisi, Faraz Ahmad, Fathia Al Murhsidi, Khalid Al Thihli, Renjith Mani (OMAN)

P06-07 | Molybden Cofactor Deficiency Type B presenting with Guillain-Barré Syndrome-like flaring of Chronic Peripheral Neuropathy, Intellectual Disability and Dysmorphism

Bülent Kara, Ayfer Sakarya Güneş, Gözde Yeşil (TURKEY)

P06-08 | Succinic Semialdehyde Dehydrogenase Deficiency – A case report

Chrysoula Michaletoú, Aikaterini Giannis, Kleio Daskalaki, Evangelia Bachlava, Elli-Maria Petrou, Georgios Stokidis, Niki Nana, Anastasia Skouma, Virginia Theodorou (GREECE)

P06-09 | Neuroimaging spectrum of GM1 Gangliosidosis with description of novel imaging phenotypes

Debasree Das, Prateek Malik, Hiren Panwala, Sniya Sudhakar, Kshitij Mankad (UNITED KINGDOM, INDIA)

P06-10 | A rare case presentation: a novel mutation in GTPBP3 gene

Dilek Cavusoglu, Muhsin Elmas, Basak Gogus, Nihal Olgac Dundar (TURKEY)

P06-11 | Fatal Neonatal Onset of Mitochondrial DNA Depletion Syndrome due to novel MPV17 gene variants in two sisters

Furene Wang, Jann Adriel Sy, Stacey Tay, Denise Goh (SINGAPORE)

P06-12 | Differential diagnosis for X-linked Adrenoleukodystrophy

Tetiana Stetsenko, **Halyna Fedushka**, Olena Savchenko, Stanislav Rebenkov (UKRAINE)

P06-13 | AXO-AAV-GM1 for the treatment of GM1 Gangliosidosis: programme overview

Gavin Corcoran, Paul Korner, Fraser Wright, Yi Mo, **Jamie Benoit**, Heather Gray-Edwards, Miguel Sena-Esteves, Douglas R. Martin (USA)

P06-14 | Quality of life in patients with Morquio A Syndrome treated in a Paediatric Neurology reference centre

Jose Cardenas, Monica Troncoso, Diane Vergara, Fernanda Balut, Veronica Saez, Isadora Ruiz, Javiera Tello, Daniela Muñoz (CHILE)

P06-15 | Neuronal Ceroid Lipofuscinoses Type 6 and impaired autophagy system

Kiwako Tsukida, Kazuhiro Muramatsu, Takahiro Ikeda, Masahide Goto, Masayo Yamazaki, Karin Kojima, Ayumi Matsumoto, Masko Nagashima, Naomichi Matsumoto, Hitoshi Osaka, Takanori Yamagata (JAPAN)

Moderated Poster Track 2 (Sessions 4-7)

P06-16 | Twenty years' follow-up of Bone Marrow Transplantation (BMT) in two patients with Adrenoleukodystrophy (ALD)

Makiko Kaga, Kotoe Sakihara, Masako Nakamura, Atsuko Gunji, Masumi Inagaki, Shunichi Kato (JAPAN)

P06-17 | The 7th Greek case of Tyrosine Hydroxylase Deficiency: even stronger evidence for a founder effect

Maria Kyriazi, Maria Nivatsi, Pinelopi Dragoumi, Efthymia Vargiami, Maria Milioudi, Marcel Verbeek, Dimitrios Zafeiriou (GREECE, NETHERLANDS)

P06-18 | Leigh Encephalopathy in patients with valine metabolism defects due to mutations in HIBCH and ECHS1 genes

Mariya Sigatullina, Heidi Baide-Marena, Laura Martí-Sánchez, Roser Pons, Javier Ortigoza-Escobar, Angel Sánchez-Montanez, Elida Vázquez, Ignacio Delgado, S. Aguilera-Albesa, Anna Ribes, L. Pollini, S. Galosi, V. Leuzzi, L. Pérez-Gay, P. Sánchez-Pintos, Cristiano Rizzo, Michela Semeraro, Diego Martinelli, Carlo Dionisi-Vici, Rafael Artuch, Belen Pérez-Dueñas (SPAIN, GREECE, ITALY)

P06-19 | Severe MTHFR Deficiency with Hyperhomocysteinemia, Demyelinating Leukodystrophy and Psycho-Motor Retardation

Mesut Güngör, Bülent Kara, Ayfer Sakarya Güneş, Gülden Gökcay (TURKEY)

P06-20 | Glucose Transporter Type 1 (Glut-1) Deficiency Syndrome: a single centre case series

Miraç Yıldırım, Ömür Babayıgit, Dilek Yalnızoğlu, Meral Topçu (TURKEY)

P06-21 | Metacromatic Leukodystrophy – Rare and serious progressive disease

Monika Kukuruzovic, Masa Malenica, Iva Separovic (CROATIA)

P06-22 | Leukodystrophy and differential diagnosis of Arylsulfatase Deficiency

Nesibe Gevher Eroglu Ertugrul, Faruk Pekgul, Bahadir Konuskan, Baris Kuskonmaz, Duygu Uckan Cetinkaya, Meral Topcu, Hatice Asuman Ozkara, Banu Anlar (TURKEY)

P06-23 | Congenital Disorder of Glycosylation (CDG) Type II associated with a SLC39A8 gene variant: description of two siblings with variable phenotype

Noelia Rivera Sanchez, Frederic Tort, Raquel Montero, Judith Armstrong, Delia Yubero, Mercedes Serrano, Antonia Ribes, Aída Ormazabal, Rafael Artuch, Angels García Cazorla, Alejandra Darling, Mar O'Callaghan (SPAIN)

P06-24 | Metachromatic Leukodystrophy: a case report of a new mutation

Atilla Ersen, **Pınar Gencpinar**, Pınar Arıcan, Sema Bozkaya Yılmaz, Ozgur Oztekin, Kadri Murad Erdogan, Nihal Olgac Dundar (TURKEY)

P06-25 | Clinical and genetic aspects of Progressive Myoclonus Epilepsy: experience in a cohort of 48 children

Ruzica Kravljanc, Maja Djordjevic, Biljana Vucetic Tadic (SERBIA)

P06-26 | Clinical characteristics and inherited metabolic or genetic aetiologies of Homocystinemia; a single centre experience

Ilknur Erol, **Şeyda Beşen**, Yasemin Özkal, Aytül Noyan (TURKEY)

P06-27 | Phenotypic spectrum of Short-Chain enoyl-CoA Hydratase-1 Deficiency

Silvia Masnada, Carlo Corbetta, Luisella Alberti, Laura Saieilli, Mario Barbarini, Paolo Bini, Chiara Doneda, Cecilia Parazzini, Luisa Chiapparini, Pierangelo Veggiotti, Davide Tonduti (ITALY)

P06-28 | Advances in the natural history of an underdiagnosed group of Neurometabolic Disorders: the International Niemann-Pick Diseases Registry (INPDR)

Flavia Viana Almeida Freitas, Amanda Beatriz Andrade, Isabela Caldas, Marc Patterson, Jackie Imre, Jim Green, Shaun Bolton, Charles Lourenco (BRAZIL, USA, UNITED KINGDOM)

Moderated Poster Track 2 (Sessions 4-7)

P06-29 | *Neuronal Ceroid Lipofuscinosis Type 2: when the "atypical" phenotypes may be the typical ones*

Isabela Caldas, Amanda Beatriz Andrade, Flavia Viana Freitas, Carolina Fischinger Moura De Souza, Andre Pessoa, Zumira Carneiro, Charles Lourenco (BRAZIL)

P06-30 | *Ethylmalonic Encephalopathy: a rare inborn error of metabolism with a unique constellation of clinic-radiological features*

Athanasis Moraitis, Euthymia Vargami, Maria Kyriazi, Penelopi Dragoumi, Athanasia Anastasiou, Maria Milioudi, Dimitrios Zafeiriou (GREECE)

P06-31 | *The natural history of renal manifestations in patients with Mitochondrial Disease*

Maria Parasyri, Per Brandstrom, Niklas Darin, Mar Tulinius, Kalliopi Sofou, MCRN investigators (SWEDEN)

P06-32 | *Progressive Demyelinating Neuropathy after hematopoietic cell transplantation in Metachromatic Leukodystrophy: a case series*

Shanice Beerepoort, Marjo S. van der Knaap, Jaap-Jan Boelens, Caroline A. Lindemans, Marianna Bugiani, Nicole I. Wolf (NETHERLANDS, USA)

P06-33 | *Neurodegeneration with Brain Iron Accumulation (NBIA) – Two cases in Bosnia and Herzegovina*

Ferija Hadzagic Catibusic, Sajra Uzicanin, Deniz Bulja, Lejla Smajic, Velma Selmanovic (BOSNIA AND HERZEGOVINA)

P06-34 | *Delays in diagnosis are associated with poor clinical outcomes in patients with Arginase 1 deficiency*

George Diaz, Nicola Longo, Gillian Bubb, Stephen Eckert, **Mark Bechter**, James Wooldridge, Lawrence Merritt (USA)

SESSION 7 MISCELLANEOUS

P07-01 | *Difficulties in the differential diagnosis of convulsions in Spinal Dysraphies: a clinical case*

Meruert Takhanova, Azamat Zhailganov, Assem Ibragimova, **Altynshash Jaxybayeva** (KAZAKHSTAN)

P07-02 | *Magnetic Resonance Spectroscopy (MRS) in Neonatal Hypoxic Ischaemic Encephalopathy (HIE)*

Chrysoula Rizava, Daniel Connolly, Lis Hoyles, Jen Larsen, Tamanna Williams, Anthony Hart (UNITED KINGDOM)

P07-03 | *Application of ketogenic diet to alleviate symptoms of Autism in children – Case series*

Iwona Zarnowska, Krystyna Mitosek-Szewczyk, Tomasz Zarnowski, Beata Chrapko, Teresa Marszałek, Zaneta Brudkowska (POLAND)

P07-04 | *Progression of neurophysiological changes in a child with Subacute Sclerosing Panencephalitis*

Abigail Lazenbury, Neeraj Bhangu, Marjorie Illingworth, David Allen, Jaspal Singh (UNITED KINGDOM)

P07-05 | *Alternating Hemiplegia of childhood – 17 years follow-up and genetic testing*

Alla Nechay, Iryna Nikolayenko (UKRAINE)

P07-06 | *Clinical and radiological features of Myelitis associated with Non-Polio Enterovirus Infections in the Western Cape Province of South Africa*

Alvin Ndondo, Ebrahim Banderker, Jo Wilmshurst (SOUTH AFRICA)

P07-07 | *Multi-disciplinary workshops improve understanding and awareness among families and professionals in Paediatric Narcolepsy*

Ashley Holt, Chetana Kallappa (UNITED KINGDOM)

Moderated Poster Track 2 (Sessions 4-7)

P07-08 | Approaches to utilising simulation-based-education in Paediatric Neurology training

Ashley Holt (UNITED KINGDOM)

P07-09 | Herpes Simplex Virus-1 as a rare aetiology of Isolated Acute Cerebellitis

Cem Paketci, Pinar Edem, Canan Okumus, Fatma Ceren Sarioglu, Erhan Bayram, Semra Hiz, Uluc Yis (TURKEY)

P07-10 | 4-year-old girl with Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS): investigation with MRI Imaging including DTI Tractography Sequence

Dimitrios Champasas, George Vartzelis, Anastasia Korona, Eleni Maria Papatesta, Georgia Amountza, Anastasia Garoufi, Ioannis Nikas (GREECE)

P07-11 | A follow-up study of clinical characteristics, disease course, disability, quality of life and psychological difficulties in children and adolescents with Migraine

Gordana Kovacevic, Dejan Stevanovic, Dragana Bogicevic, Dimitrije Nikolic, Slavica Ostojic, Biljana Vucetic Tadic, Blazo Nikolic, Ivana Bosiocic, Nikola Ivancevic, Kristina Jovanovic, Janko Samardzic, Jasna Jancic (SERBIA)

P07-12 | Quality-of-life evaluation of healthy siblings of children with Cerebral Palsy and Epilepsy

Meltem Dinleyici, **Kursat Bora Carman**, Coskun Yarar, Aysu Duyan Camurdan, Figen Sahin Dagli (TURKEY)

P07-13 | Arachnoid Cyst in children: the relationship of clinical features and neuroanatomical location

Kursat Bora Carman, Güldeniz Coskuner, Suzan Saylisoy, Coskun Yarar (TURKEY)

P07-14 | The clinical profile and outcome of children with Tuberous Sclerosis in Durban South Africa

Lionel Nyamurenje, Lawrence Mubaiwa (SOUTH AFRICA)

P07-15 | Recurrent Hemiplegic Migraine induced by Exertion: a singular presentation of en Coup de Sabre Scleroderma

Rita Martins, **Sofia Quintas**, João Ferreira, Tiago Proença Santos, Joana Coelho, Freitas João, António Levy (PORTUGAL)

P07-16 | Meningococcal Septicaemia presenting as Acute Brachial Plexus Neuritis

Sadaf Ahsan, Ala Fadilah, Chris Rittey (UNITED KINGDOM)

P07-17 | Unilateral Facial Erythema following food chewing

Samar Almuntaser, Maysa Saleh, Gururoj Aithala (UNITED ARAB EMIRATES)

P07-18 | Acute Ataxia in children: common causes and yield of diagnostic work-up in the post-Varicella vaccine era

Sarit Ravid, Aharon Schif, Imad Kassis, Elena Segal (ISRAEL)

P07-19 | Cosyntropin mediates Melanocortin Microglial Inflammation and improves outcomes in rodent TBI

Stephen Ashwal, Lorraine Siebold, Christopher Wilson, Brenda Bartrik-Olson, Johnny Figueroa, Julio Vega-Torres, Beatriz Tone, Barbara Holshouser (USA)

P07-20 | Critical role for the thalamus, corpus callosum and basal ganglia in paediatric TBI: results of a longitudinal imaging study

Stephen Ashwal, Jamie Pivonka-Jones, Joy Nichols, Udo Oyoyo, Bartrik-Olson Brenda, Karen Tong, Barbara Holshouser (USA)

P07-21 | A paediatric case of Lyme Neuroborreliosis presenting with abdominal Radiculitis

Ivan Fiorito, **Thomas Foiadelli**, Salvatore Savasta (ITALY)

Moderated Poster Track 2 (Sessions 4-7)

P07-22 | *Scurvy. An autistic boy refuses to walk: the comeback of an ancient disease*

Tinneke Stals, Rosalind Verheije, Lieve Lagae, Gunnar Buyse, Nathalie Goemans, Liesbeth De Waele, Katrien Jansen, Els Ortibus (BELGIUM)

P07-23 | *Critical illness Polyneuropathy and Myopathy in children: case reports and literature review*

Tomoko Uchida, Ktsunori Fujii, Tadashi Shiohama, Hironobu Kobayashi, Naoki Saito, Naoki Shimojo (JAPAN)

P07-24 | *Scoliosis in children and young people with Ataxia Telangiectasia (A-T)*

William Whitehouse, Jeyanthi Rangaraj, Ouliana Piagioti, Min Ong, Masood Shafafy, Mohnish Suri (UNITED KINGDOM)

P07-25 | *Multidisciplinary management of Neurofibromatosis Type 1 (NF-1): the experience from two (2) paediatric hospitals in Greece*

Eleftheria Kokkinou, Dionysia Gkougka, Christina Pourlou, Kleoniki Roka, Alexis Alexopoulos, Efthymia Tsina, Ioannis Nikas, Panagiotis Krallis, Lambrini Nasi, Ioanna Thanopoulou, Evangelia Makrygianni, Maria Tzetzis, Maria Tsipi, Irene Tsoutsou, Konstantina Kosma, Sofia Kitsou, Eleni Fryrisa, Evangelos Paraskevoulakos, Antonis Kattamis, Roser Pons, Charalambos Kotsalis (GREECE)

P07-26 | *Validation of the use of WhatsApp as a method of communication with families*

Alfonso Amado Puentes, Noa Villar, Sara Pereiro (SPAIN)

P07-27 | *Children and adolescents with disabilities and the first three years of experience from the first hospice for children and adolescents in Denmark*

Charlotte Pedersen, Susan Cawley (DENMARK)

P07-28 | *Idiopathic Intracranial Hypertension: a paediatric case series*

Joana Coelho, Tiago Proença dos Santos, Bárbara Marques, Sofia Quintas, António Levy (PORTUGAL)

P07-29 | *Brainstem beaklike. A case report*

Noelia Rivera Sanchez, Mónica Rebollo Polo, Verónica Delgadillo, Xènia Alonso Curco (SPAIN)

P07-30 | *Influence of pineal gland cyst on hypothalamic-pituitary hormones in children*

Tadeja Hostnik, Rok Kučan, Primož Kotnik, Mirjana Perković Benedik, Damjan Osredkar (SLOVENIA)

P07-31 | *Characterization of speech and voice abnormalities in a cohort of GLUT1DS patients*

Leticia Pias, Alejandra Darling, Núria Montagut Colomer, Rosanna Mari Vico, Marta Gil González, Roser Colomer Roura, Fanny Mochel, Roser Pons, Angels García-Cazorla (SPAIN, FRANCE, GREECE)

P07-32 | *A case of cutis Verticis Gyra associated with intellectual disability and Epilepsy*

F. Mujgan Sonmez, Seval Erpolat (TURKEY)

P07-33 | *Tuberous Sclerosis: evaluation of 115 patients, single centre study, Turkey*

Aslıhan Akdemir, Hakan Gümüş, Mehmet Canpolat, Sefer Kumandas, Hüseyin Per (TURKEY)

P07-34 | *Neurofibromatosis Type 1: evaluation of 124 patients, single centre study, Turkey*

Aslıhan Akdemir, Hakan Gümüş, Mehmet Canpolat, Sefer Kumandas, Hüseyin Per (TURKEY)

P07-35 | *Sturge-Weber Syndrome: evaluation of 15 patients, single centre study, Turkey*

Aslıhan Akdemir, Hakan Gümüş, Mehmet Canpolat, Sefer Kumandas, Hüseyin Per (TURKEY)

Moderated Poster Track 3 (Sessions 8-11)

SESSION 8 EPILEPSY: MISCELLANEOUS, NEUROREHABILITATION

P08-01 | Significant hidden socio-emotional problems in children with neurological disorders revealed by sentence completion test

Anneli Kolk, Marianne Saard, Christen Kööp, Lisanna Pertens, Kirsi Sepp (ESTONIA)

P08-02 | Social scenarios for virtual reality platform for remediation of Social Communication Disorder in children

Anneli Kolk, Marianne Saard, Lisanna Pertens, Kirsi Sepp, Liina Reinart, Christen Kööp (ESTONIA)

P08-03 | Continuous EEG monitoring in cases with Acute Non-Traumatic or Non-Opera Traumatic Encephalopathy

Ozlem Balci, Hepsen Mine Serin, Erdem Simsek, Seda Kanmaz, İpek Cetin, Pinar Yazıcı, Bulent Karapınar, Gul Aktan, Hasan Tekgül, Sanem Yılmaz, Sarenur Gökben (TURKEY)

P08-04 | Radial Nerve Injury in the newborn. Case series

Adrian Ioan Toma (ROMANIA)

P08-05 | Difficulties in diagnosing of SSPE in Kazakhstan presenting at three cases

Altynshash Jaxybayeva, Assel Abilkhadirova, Alissa Nauryzbayeva, Zhazira Khaldarova (KAZAKHSTAN)

P08-06 | Can neonatal staff site EEG leads in the correct location? A pilot study

Anthony Hart, James Alix (UNITED KINGDOM)

P08-07 | What Epilepsies are seen following moderate to severe Neonatal Hypoxic-Ischaemic Encephalopathy (HIE)

Anthony Hart, Coronwy Hughes, James Alix (UNITED KINGDOM)

P08-08 | Role of the deprivation of liberty process in Paediatric Neurorehabilitation practice

Archana Murugan, Megan Eve, Peta M Sharples (UNITED KINGDOM)

P08-09 | Infantile spasms following Acquired Brain Injury – A tertiary Neuro-Rehabilitation centre experience

Archana Murugan, Christopher J. Butler, Peta M. Sharples (UNITED KINGDOM)

P08-10 | Management of Neonatal Seizures in a Level 2 Neonatal Unit in the United Kingdom

Ashley Holt (UNITED KINGDOM)

P08-11 | Correlations among functional classification systems in Cerebral Palsy: a study using the Surveillance of Cerebral Palsy in Europe (SCPE) Database

Sefa Uner, Ceren Gunbey, Nesibe Gevher Erogul Ertugrul, Merve Tuncdemir, Cemil Ozal, Ozge Cankaya, Kubra Seyhan, Kivanc Delioglu, Mintaze Kerem Günel, Banu Anlar (TURKEY)

P08-12 | Two phase 1 healthy volunteer trials investigating the potential effects of CYP3A4 and CYP2C19 inhibition or induction on Cannabidiol (CBD) Pharmacokinetics

David Critchley, Gilmour Morrison, Joris Berwaerts, Julie Crockett, Sam Greenwood, Bola Tayo (UNITED KINGDOM)

P08-13 | The effect of meal composition, milk, and alcohol on Cannabidiol (CBD) exposure and safety after a single dose of CBD oral solution in healthy adults: results of a Phase 1, randomized, crossover, Pharmacokinetic (PK) trial

David Critchley, Bola Tayo, Joris Berwaerts, Julie Crockett, Gilmour Morrison (UNITED KINGDOM, USA)

Moderated Poster Track 3 (Sessions 8-11)

P08-14 | Evaluation of the electroclinical phenotype in Pallister-Killian Syndrome: preliminary results

Emilia Ricci, Rocco Bonfatti, Alessandro Rocca, Giacomo Sperti, Guido Cocchi, Valeria Cagnazzo, Aglaia Vignoli, Duccio Maria Cordelli (ITALY)

P08-15 | Idiopathic Infantile Spasm with a good outcome

Esra Serdaroglu (TURKEY)

P08-16 | Febrile Infection-Related Epilepsy Syndrome (FIRES): clinical presentation, treatment and outcome in a series of patients

Fatima Delgado Ledesma, Estibaliz Barredo Valderrama, María Concepción Miranda Herrero, María Vázquez López, Pedro De Castro De Castro (SPAIN)

P08-17 | Longitudinal trajectories of self-reported depressive symptoms in children with Epilepsy

Gabriel Ronen, Tea Rosic, David Streiner (CANADA)

P08-18 | Febrile Infection-Related Epileptic Syndrome (FIRES) case report presentation: a challenging medical event with ominous outcome

Ilias Georgiadis, Eftychia-Maria Kontouri, Maria Sarigianni, Anastasia Anastasiou-Katsiardani (GREECE)

P08-19 | The course of Epilepsy with continuous spike-waves during sleep in children with Cerebral Palsy

Ina Kozyrava, Sviatlana Kulikova, Sergei Likhachev, Tatyana Svinkovskaja, Igor Zaitsev, Sviatlana Belaja, Marina Savchenko (BELARUS)

P08-20 | Transient elevation of TSH following seizures in children

Ji Yoon Han, Bi Na Kim (REPUBLIC OF KOREA)

P08-21 | Clinical case of boy with Hypothalamic Hamartoma and seizures

Kleio Daskalaki, Aikaterini Giannisi, Chrysoula Michaletou, Georgios Stokidis, Niki Nana, Evangelia Bachlava, Elli-Maria Petrou, Anna Christodoulaki, Anastasia Nalbanti, Marina Katsalouli, Virginia Theodorou (GREECE)

P08-22 | A novel method to analyse and monitor walking capacity in children with Cerebral Palsy, to identify response after treatment

Kyriakos Martakis, Christina Stark, Oliver Semler, Ibrahim Duran, Eckhard Schoenau (GERMANY)

P08-23 | Dyke-Davidoff-Masson Syndrome: a case report in a Filipino male adolescent

Lalaine Villaflor-Oida, Maria Antonia Aurora Valencia, Alejandro Bimbo Diaz (PHILIPPINES)

P08-24 | First report of ictal self-harm in a child with a Generalised Seizure Disorder with Absences and Eyelid Myoclonus

Leena Mewasingh, Christina Korley, Matthew Sparkes, Sushma Goyal (UNITED KINGDOM)

P08-25 | Influence of cognitive and behavioural comorbidities on the level of Depression and Anxiety among children with Epilepsy

Dana Buršíková Brabcová, Jiří Kohout, Anežka Bělohlávková, **Matyáš Ebel**, Jitka Rokytová, Pavel Kršek (CZECH REPUBLIC)

P08-26 | A new guideline on the management of Spasticity in Hereditary Spastic Paraparesis

Rushna Raza, Raj Lodh (UNITED KINGDOM)

P08-27 | A child with Persistent Epileptic Seizures and Long QT diagnosed with Tuberous Sclerosis Complex, after the findings in brain MRI

Stella Mouskou, Kallirroi Kamizi, Aikaterini Koumparelou, Maria Lyra, Ioannis Nikas, Georgios Servos, Nikos Marinakis, Danai Veltra, Sotiria Mastroianni, Anastasia Korona, George Vartzelis, Vasiliki Houriara, Jan Traeger-Synodinos, Efstathia Katsarou-Pectasides, Konstantinos Voudris (GREECE)

Moderated Poster Track 3 (Sessions 8-11)

P08-28 | Features of Autosomal Dominant Nocturnal Frontal Lobe Epilepsy in patients with mutations in the KCNT1 gene

Sviatlana Kulikova, Sergey Likhachev, Inna Kozyreva, Anastasia Kuznecova (BELARUS)

P08-29 | The first case of identification of KCNMA1 mutations in a child with a clinical diagnosis of juvenile Absence Epilepsy in Ukraine

Tetiana Stetsenko (UKRAINE)

P08-30 | Use of the Rehabilitation Complexity Scale to assess dependency and rehabilitation complexity in children receiving in-patient Neurorehabilitation at a Regional Neuroscience Centre (RNSC)

Todd Smallbone, Biju Hameed, Robert Spaull, C. Reyes Payeras, Eve Megan, Ingram Wright, Peta Sharples (UNITED KINGDOM)

P08-31 | Treatment Resistant Epilepsy and Spasticity in Infantile Ceroid Lipofuscinosis (INCL): a case report

Annika Tihveräinen, Jan Olme (FINLAND)

P08-32 | Children's adherence to antiepileptic medication: does socioeconomic status play a role? A systematic review

Andrea Nahum, Jonathan Searle (UNITED KINGDOM)

P08-33 | Periodical Lateralized Epileptiform Discharges (PLEDs) with Hippocampal Atrophy in a child with past medical history of Benign Focal Epilepsy

Elli-Maria Petrou, Aikaterini Giannisi, Kleio Daskalaki, Evangelia Bachlava, Chrysoula Michaletou, Georgios Stokidis, Niki Nana, Anna Christodoulaki, Anastasia Nalbanti, Dimitrios Verganelakis, Maria Chasiotou, Virginia Theodorou (GREECE)

P08-34 | Motor Dysfunction in Neurofibromatosis Type 1 with normal neurological examination: a muscular problem?

Gokce Gurler, Hira Altunbuker, Sinem Sel, Ozge Cankaya, Mintaze Kerem Günel, Nesibe Gevher Eroglu Ertugrul, Banu Anlar (TURKEY)

P08-35 | Comprehensive approach to Paediatric Rehabilitation: creating a structured model of Neurorehab for social competence training in children

Marianne Saard, Anneli Kolk, Lisanna Pertens, Christen Kööp, Kirsi Sepp, Liina Reinart (ESTONIA)

P08-36 | Drug Resistant Epilepsy in infant with GRIN2A, SPTAN1 and SCN2A mutations and probably Congenital Cytomegalovirus Infection

Dorota Dunin-Wasowicz, Dorota Domanska-Pakiela, Katarzyna Tomaszek, Elzbieta Ciara (POLAND)

P08-37 | DAT Questionnaire – Early detection of disorders associated with Dravet Syndrome (DS)

Igor Prpic, Inge Vlasic-Cicvaric, Jelena Radic Nisevic, Ivana Kolic, Nusa Naglic (CROATIA)

P08-38 | Transition in Epilepsy – Croatian experience

Masa Malenica, Igor Prpic, Inge Vlasic-Cicvaric, Ana Sruk, Iris Zavoreo, Nevia Novak, Jelena Radic-Nisevic, Monika Kukuruzovic, Iva Separovic, Ivana Kolić, Marija Oreskovic (CROATIA)

Moderated Poster Track 3 (Sessions 8-11)

SESSION 9 MOVEMENT DISORDERS

P09-01 | SPG15 presenting as Early Onset Parkinsonism in a teenager: a case of Levodopa responsiveness
Amanda Beatriz Andrade, Isabela Caldas, Flávia Viana Almeida Freitas, Laura Vagnini, Filippo Santorelli, Charles Lourenco
(BRAZIL, ITALY)

P09-02 | The applicability of the Scale for Assessment and Rating of Ataxia (SARA) in toddlers
Sjoukje Polet, Sahar Hbrahimgel, Anna Tadema, Marloes van den Berg, Rick Brandsma, Marieke Kuiper, **Deborah Sival**
(NETHERLANDS)

P09-03 | Stabilisation of Progressive Dystonia with sequential STN DBS and ITB in a case of Pantothenate Kinase associated Neurodegeneration
Lisa Myers, Simon Paget, Russell Dale, Neil Mahant, Hugo Sampaio, Maria Kyriagis, **Shekeeb Mohammad** (AUSTRALIA)

P09-04 | A single centre experience of diagnostic yield in Ataxia in childhood
Ala Fadilah, Peter S. Baxter, Santosh R. Mordekar (UNITED KINGDOM)

P09-05 | Tone abnormalities and feeding difficulties in Allan-Herndon-Dudley Syndrome (AHDS): recognition of the natural history
Ala Fadilah, Emily Davies, Paul Dimitri, Santosh R. Mordekar (UNITED KINGDOM)

P09-06 | Functional parameter measurements in Ataxia Telangiectasia – A cross sectional study
Efrat Shenhod, Bruria Ben Zeev, Ifat Sarouk, Gali Heimer, **Andreea Nissenkorn** (ISRAEL)

P09-07 | Secondary Enuresis and urological manifestations in children with Ataxia Telangiectasia
Andreea Nissenkorn, Tomer Erlich, Dorit Zilberman, Ifat Sarouk, Alexander Krauthammer, Noam Kitrey, Gali Heimer, Bruria Ben Zeev, Yoram Mor (ISRAEL)

P09-08 | Expanding the phenotypic spectrum of CACNA1 gene mutation
Bola Kazeem, Manali Chitre, S.J. Mehta, Bina Mukhtyar (UNITED KINGDOM)

P09-09 | AADC Deficiency in Latin-America: improving diagnosis and awareness of a rare neurometabolic disease
Charles Lourenco, Juliana Cardoso, Kimbali Ventureli, Rayssa Santos, Jacqueline Fonseca, Laura Vagnini, Jose Francisco Franco (BRAZIL)

P09-10 | Systematic Movement Disorder evaluation including EMG assessment provides increased insight in children with Dyskinetic Cerebral Palsy
Jakob Lorentzen, Alfred Peter Born, Christian Svane, Christian Forman, Bjarne Laursen, Annika Reynberg Langkilde, Peter Uldall, Jens Bo Nielsen, **Christina Engel Hoei-Hansen** (DENMARK)

P09-11 | Different faces of Movement Disorders in children
Coskun Yasar, Kursat Bora Carman, Sultan Durmus Aydogdu, Ozan Kocak, Gonca Kilic Yildirim, Emre Kaplan, Sibel Gurlevik Lacinel, Eylem Kiral, Sevgi Yimenicioglu, Ilhan Isik, Arzu Ekici, Serhat Ozkan, Gurkan Bozan, Oguz Cilingir, Serdar Ceylaner, Ayten Yakut (TURKEY)

P09-12 | Spectrum of Alternating Hemiplegia of childhood – Experience of our clinic
Cristina Andreea Minca, Oana Tarta-Arsene, Diana Barca (ROMANIA)

P09-13 | Intravenous Immunoglobulin therapy for childhood Fisher Syndrome
Daisuke Sawada, Katsunori Fujii, Taku Omata, Masakazu Honda, Akira Ohtake, Kensuke Fujishiro, Yukiko Iwase, Maiko Suyama, Tomoko Uchida, Tadashi Shiohama, Hideki Uchikawa, Naoki Shimojo (JAPAN)

Moderated Poster Track 3 (Sessions 8-11)

P09-14 | Cognition and emotion regulation in Early Onset Ataxia

Maraike Coenen, **Deborah Sival**, Rick Brandsma, Marina Tijssen, Jacoba Spikman (NETHERLANDS)

P09-15 | Can we phenotypically distinguish Early Onset Ataxia from Developmental Coordination Disorders?

Tinka Lawerman, Rick Brandsma, Natasha Maurits, Octavio Martinez-Manzanera, Roelineke Lunsing, Oebele Brouwer, Berry Kremer, **Deborah Sival** (NETHERLANDS)

P09-16 | Exergame training in Early Onset Ataxia patients

Rick Brandsma, Ilse Ganzevoort, Marloes van den Berg, Zeus Dominguez Vega, Dorothee Jelsma, Marina Schoemaker, Natasha Maurits, Claudine Lamoth, **Deborah Sival** (NETHERLANDS)

P09-17 | Usefulness of MRI Classification System (MRICS) to determine the aetiology of Cerebral Palsy

Eszter Nagy, Zsuzsanna Herbert, István Péter, Eszter Csorba, Andrea Skobrák, Nelli Farkas, Katalin Hollódy (HUNGARY)

P09-18 | Application of next-generation sequencing in the identification of genes associated with paediatric Movement Disorders

Federica Graziola, Giacomo Garone, Luca Bosco, Fabiana Stregapede, Enrico Bertini, Paolo Curatolo, Federico Vigevano, Lorena Travaglini, Alessandro Capuano (ITALY)

P09-19 | Audit of a tertiary centre experience with symptoms (Tics, Tourettes, OCD) within the PANDAS spectrum

Laura Royce, Fenella Kirkham (UNITED KINGDOM)

P09-20 | Genetic causes of Congenital Mirror Movements associated with brain malformations

Lubov Blumkin, Zvi Leibovitz, Keren Yosovich, Itay Zelcer, Ayelet Zerem, Liat Gindes, Dorit Lev, Tally Sagie (ISRAEL)

P09-21 | Coexistence of mutations in FBXO7 and DLD causes a Complex Neurodegenerative Disorder with clinical and neuro-radiological overlapping with NBIA

Marta Correa-Vela, Wiktoria Torbé, Vicenzo Lupo, Paula Sancho, Anna Marcé-Grau, Alejandra Darling, Sandra Fernández-Rodríguez, Alison Jenkins, Cristina Tello, Laura Ramírez, Carmen Espinós, María J. Sobrido, Belén Pérez-Dueñas (SPAIN)

P09-22 | Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy, childhood-onset; NADGP: a case report

Mehmet Canpolat, Hamit Acer, Sevgi Çıraklı, Murat Erdoğan, Sefer Kumandaş (TURKEY)

P09-23 | ADCY5-related Dyskinesia: report of 2 patients

Mireia Vázquez-Piqueras, Delia Yubero, Alberto García-Oguiza, Jaume Campistol, Juan Darío Ortigoza-Escobar (SPAIN)

P09-24 | Response of Movement Disorder to Deep Brain Stimulation in patients with variants in GNAO1

Mireia Vázquez-Piqueras, Jordi Munchart, Cristina Cáceres-Marzal, Alejandra Darling, Santiago Candela, Alejandra Climent, Jordi Rumià, Juan Darío Ortigoza-Escobar (SPAIN)

P09-25 | Clinical description of Ataxia in Ataxia-Telangiectasia (Louis-Bar Syndrome)

Nesibe Gevher Eroglu Ertugrul, Sefa Unes, Merve Tuncdemir, Busranur Cavdarli, Mintaze Kerem Gunel, Banu Anlar (TURKEY)

P09-26 | Variant Ataxia-Telangiectasia in a child presenting with Laryngeal Dystonia

Pinar Arican, Nihal Olgac Dundar, Ozgur Kirbiyik, Dilek Cavusoglu, Sema Bozkaya Yilmaz, Pinar Gencpinar (TURKEY)

P09-27 | A case of Spastic Paraplegia-15 with a novel pathogenic variant in ZFYVE26 Gene

Taha Resid Ozdemir, **Pinar Gencpinar**, Pinar Arican, Ozgur Oztekin, Nihal Olgac Dundar, Berk Ozyilmaz (TURKEY)

P09-28 | Focal Cervical Dystonia in Stickler Syndrome – Supporting the role of visuospatial processing contributing to dysfunction of the head neural generator

Saraswathy Sabanathan, Philip Alexander, Sarah Bowdin, Eric Ezra, Brinda Muthusamy, Richard Brown, **Pooja Harijan** (UNITED KINGDOM)

Moderated Poster Track 3 (Sessions 8-11)

P09-29 | High doses of Intrathecal Baclofen in the treatment of Severe Generalized Dystonia in a patient with Pantothenate Kinase-associated Neurodegeneration

Sviatlana Kulikova, Sergey Likhachev, July Rushkevich, Vladimir Alexeevets, Alexandr Korene (BELARUS)

P09-30 | Development of treatment strategies for MPAN, a NBIA disorder

Enrica Zanuttigh, Caterina Terrile, Ejona Rusha, Holger Prokisch, Thomas Klopstock, Thomas Meitinger, Arcangela Iuso (GERMANY)

P09-31 | Bridging regimen of Enteral to Transdermal Clonidine for management of paediatric Movement Disorders

Guo Yong Lim, Ling Ying Tan, Zhi Min Ng, Tong Hong Yeo (SINGAPORE)

P09-32 | Early-Onset Parkinsonism

Daniela Munoz, Monica Troncoso, Valentina Naranjo, Jose Hidalgo, Isadora Ruiz, Paola Santander, Susana Lara, Veronica Saez, Javiera Tello, Paulina Alid, Andres Barrios (CHILE)

P09-33 | Classic Ataxia Telangiectasia beyond the age of 30 years

Nienke Van Os, Corry Weemaes, Marcel van Deuren, Bart van de Warrenburg, Michèl Willemse (NETHERLANDS)

SESSION 10 NEURODEVELOPMENTAL

P10-01 | Electroencephalographic changes in children with Speech Disorder after late prematurity in neonatal period

Tetyana Yanina, Larisa Fedorova, Olga Rybalko (RUSSIAN FEDERATION)

P10-02 | Neurodevelopmental consequences of Traumatic Brain Injury in children

Seyedeh Hoda Khatib-Masjedi, Ali Mohammad Pourbagher-Shahri, Ali Rajabpour-Sanati (IRAN)

P10-03 | Targeted study of the executive functions in Paediatric Stroke finalized at telematics cognitive training

Alice Zanetti, Carlotta Rivella, Paola Viterbori, Valentina De Franchis, Arianna Masiello, Anna Ronchetti, Marta Bertamino, Giulia Amico, Mariasavina Severino, Sara Signa, Giulia Prato, Paolo Moretti (ITALY)

P10-04 | Indole Tryptophan Metabolism and Cytokine S100B in children with ADHD: daily fluctuations, response to Methylphenidate and interrelationship with depressive symptomatology

Luisa Fernández-López, Ana Checa-Ros, Antonio Molina-Carballo, Isabel Cubero-Millán, Irene Machado-Casas, Antonio Muñoz-Hoyos (SPAIN, UNITED KINGDOM)

P10-05 | A rare case of Global Developmental Delay – Gillespie Syndrome

Andrei-Dan Marinescu, Cosma-Iuliu Bacos, Cristina-Andreea Minca, Diana-Gabriel Barca (ROMANIA)

P10-06 | Paroxysmal Tonic Upgaze – A multifactorial disorder responsive to Carboanhydrase inhibition

Annegret Quade, Anne Thiel, Ingo Kurth, Manuel Holtgrewe, Miriam Elbracht, Dieter Beule, Ute Scholl, Martin Häusler (GERMANY)

P10-07 | Sleep quality in children with Attention Deficit Hyperactivity Disorder (ADHD) and Sensory Modulation Difficulties (SMD)

Aviva Mimouni Bloch, Hagar Offek, Sara Rosenblum, Edith Posener, Riva Tauman, Batya Engel-Yeger (ISRAEL)

P10-08 | Magnetic Resonance Imaging findings in children with Developmental Delay from resource limited areas

Birendra Rai, Apurba Ghosh, Dipankar Das (UNITED KINGDOM, INDIA)

Moderated Poster Track 3 (Sessions 8-11)

P10-09 | Neonatal Hypoxic Ischemic Encephalopathy, seizures and neurological outcome: insight from the EEG spectral analysis

Cristina Forest, Valeria Spagnolo, Mario Ermani, Elisa Cainelli, Agnese Suppiej (ITALY)

P10-10 | Motor outcome after Therapeutic Hypothermia in infants with Hypoxic-Ischaemic Encephalopathy

Marieke Kuiper, Linda Meiners, Elisabeth Chandler, Rick Brandsma, Arend Bos, Henk ter Horst, **Deborah Sival** (NETHERLANDS)

P10-11 | Cognitive Visual Dysfunctions in children with Autism Spectrum Disorders and other developmental disabilities

Young Hoon Kim, **Eu Gene Park** (REPUBLIC OF KOREA)

P10-12 | Correlation between weak clinical signs of Epilepsy and the findings of the first EEG record in a population of patients with diagnoses of Epilepsy and ASD

Luis Marhuenda-Bermejo, Magdalena Garcia-Navarro, Patricia Andreo-Lillo, Lorena Pastor-Ferrández, **Francisco Carratalá-Marco** (SPAIN)

P10-13 | Prevalence of non-benign and probably non-benign CGH-Array CNV among a referral population of ASD children

Francisco Carratalá-Marco, Patricia Andreo-Lillo, Lorena Pastor-Ferrández, Andrea Huertas-Viudes, Francisco Galán-Sánchez, Mercedes Juste-Ruiz (SPAIN)

P10-14 | Universal newborn hearing screening in a mother-child public hospital in South Africa

Jacqueline Kim Bezuidenhout, Katijah Khoza-Shangase, Tim De Maayer, Renate Strehlau (SOUTH AFRICA)

P10-15 | Study of the efficacy of behavioural parent training in the treatment of Egyptian school-aged children with Attention Deficit Hyperactivity Disorder

Marwa Abd Elmaksoud, Hanan Azouz, Faten Abdel Latif, Tarek Omar Omar, Mona Khalil (EGYPT)

P10-16 | Visual perception processing in children with reading disabilities and who are born with very low birth weight

Miho Fukui, Shuichi Shimakawa, Hikaru Kitahara, Motoko Ogino, Tomohito Okumura, Mika Otsuki, Eiji Wakamiya (JAPAN)

P10-17 | Study on cognitive profiles of patients with 22q11.2 Deletion Syndrome: comparison with the findings of Williams Syndrome

Miho Nakamura, Fumio Hayakawa, Ryusuke Kakigi (JAPAN)

P10-18 | Economic burden of care and treatment options for patients with Rett Syndrome: two systematic literature reviews

Omar Dabbous, Vanessa Taieb, Emna Abdennadher, Meryem Bouchemi, Justyna Chorząż, Katarzyna Borkowska, Veneta Georgieva, Bryan E. McGill, Thomas A. Macek, Benit Maru, Ramesh Arjunji (USA, UNITED KINGDOM, TUNISIA, POLAND, BULGARIA)

P10-19 | Understanding early relationship between Autism Spectrum Disorder, Developmental Delay and Epilepsy in infants with Tuberous Sclerosis Complex: preliminary results from the EPISTOP Project

Romina Moavero, Arianna Benvenuto, Martina Siracusano, Leonardo Emberti Gialloreti, Eleonora Aronica, Marta Feucht, Anna Jansen, Floor Jansen, Sergiusz Jozwiak, Katarzyna Kotulska-Jozwiak, Pavel Krsek, David Kwiatkowski, Rima Nababout, Kate Riney, Bernard Weschke, Paolo Curatolo (ITALY, NETHERLANDS, AUSTRIA, BELGIUM, POLAND, CZECH REPUBLIC, USA, FRANCE, AUSTRALIA, GERMANY)

P10-20 | Novel gene mutation of Molybdenum Cofactor Deficiency

Samar Almuntaser, Maysa Saleh, Pawan Kayshape (UNITED ARAB EMIRATES)

Moderated Poster Track 3 (Sessions 8-11)

P10-21 | Missense variant in the ASXL2 gene in a teenage girl with Developmental Delay, Hypotonia and Bilateral Ptosis: variant of Sashi Pena Syndrome?

Sarah Buts, Patrick Verloo, Bjorn Menten (BELGIUM)

P10-22 | The Role of the arcuate fasciculus depending on hemisphere in children with Developmental Delay: a preliminary study

Sehee Kim, Kyung Eun Nam, Ah-Ra Cho, Joo Hyun Park (REPUBLIC OF KOREA)

P10-23 | DiGeorge Syndrome presenting with seizures

Seyedeh-Hoda Khatib-Masjedi, Samaneh Noroozi-Asl, Ali Rajabpour-Sanati (IRAN)

P10-24 | Epidemiology of Intellectual Disability (ID) in a population-based cohort of patients with Cerebral Palsy (CP)

Sotiria Mastroianni, Irini Nikaina, Marianna Petra, Maria Koutsaki, Maria Pyrgelli, Helen Strataki, Nikoletta Smyrni, Helen Bouza, Margarita Tzaki, George Damianos, Zoi Dalivigka, Stella Mouskou, Konstantinos Voudris, Argirios Dinopoulos, Helen Skouteli, Efstathia Katsarou, Antigone Papavasiliou (GREECE, USA)

P10-25 | Preschool screening of children aged 5 years to evaluate Neurodevelopmental Disorders in a rural city of Japan

Toshihiro Suzuki (JAPAN)

P10-26 | Pilot study to investigate the microstructural brain changes after taking Methylphenidate in children with Attention-Deficit/Hyperactivity Disorder (ADHD)

Winnie Wan-Yee Tso, Edward Hui, Vince Vardhanabhuti, Brian Ip, **Cheuk Wing Fung** (HONG KONG)

P10-27 | Cerebellar disruption at term equivalent age and motor impairment at 3 years of age in preterm infants with mild intraventricular haemorrhage: quantitative assessment using Diffusion Tensor Imaging

Satoshi Sakaue, Tatsuji Hasegawa, Eisuke Ichise, Akari Takai, Yui Zen, Michiko Yoshida, Takenori Tozawa, Tomohiro Chiyonobu, Masafumi Morimoto (JAPAN)

P10-28 | The role of Topiramate in the management of childhood Tourette's Syndrome: a case series

Helena Fawdry, Andrew Curran (UNITED KINGDOM)

P10-29 | Biallelic Neurofascin variants affect paranodal axoglial junctions causing neurodevelopmental impairment and central and peripheral demyelination

Stephanie Efthymiou, Vincenzo Salpietro, Jerome Devaux, Maria Nolano, Henry Houlden (UNITED KINGDOM, FRANCE, ITALY)

P10-30 | Novel compound heterozygous STN1 variants can cause Coats Plus Syndrome

Tanvi Acharya, Alasdair Parker, Yanick Crow (UNITED KINGDOM)

P10-31 | Evaluation of the malnutrition risk and the factors affecting it by applying the STRONGkids Scale in Pediatric Neurology service using the STRONGkids Scale

Satı Hülya Türkmen, **Aycan Unalp**, Selvinaz Edizer, Çiğdem Ecevit (TURKEY)

P10-32 | Nuchal cord prevalence and relation with neurodevelopmental outcome at the age of one years through the five years

Edina Karabeg, Adi Karabeg, Enes Karabeg, Jasmina Rešić-Karara, Biserka Rešić (BOSNIA AND HERZEGOVINA, NORWAY, CROATIA)

Moderated Poster Track 3 (Sessions 8-11)

SESSION 11 NEUROMUSCULAR

P11-01 | Long term analysis of the rate of respiratory function decline in patients with Duchenne Muscular Dystrophy (DMD) in a real-world setting: the SYROS Study

Laurent Servais, Hank Mayer, Craig McDonald, Chora Straathof, Ulrike Schara, Thomas Voit, Eugenio Mercuri, G. Buyse on behalf of the SYROS investigators (FRANCE, USA, NETHERLANDS, GERMANY, UNITED KINGDOM, ITALY, BELGIUM)

P11-02 | Consistent long-term effect of Idebenone on the rate of respiratory function decline in advanced patients with Duchenne Muscular Dystrophy (DMD)

Laurent Servais, Oscar Henry Mayer, Craig McDonald, Thomas Voit, Eugenio Mercuri, G. Buyse on behalf of the SYROS investigators (FRANCE, USA, UNITED KINGDOM, ITALY, BELGIUM)

P11-03 | Intraneuronal Perineuroma: an unusual cause of Left Sciatic Neuropathy in childhood

Ala Fadilah, Archana V. Desurkar, Grainne Bourke, Azzam Ismail, Min T. Ong (UNITED KINGDOM)

P11-04 | Cardiac Troponin T (cTnT) as a highly sensitive parameter for Spinal Muscular Atrophy (SMA) in a floppy infant

Alexandra Ille, Andre van Egmond-Fröhlich, Simone Weiss, Magdalena Gosk-Tomek, Anna Kellersmann, Manuela Födinger, Sonja Peithner, Günther Bernert (AUSTRIA)

P11-05 | Clinical and genetic characteristic of Dystrophinopathy in children in Kazakhstan

Bakhytkul Myrzaliyeva, Marzhan Lepessova, Altynshash Jaxybayeva (KAZAKHSTAN)

P11-06 | Compensation strategies used to move a filled glass to the mouth in Duchenne Muscular Dystrophy

Karin Naarding, Linda van Schaik-Bank, Mariska Janssen, Menno Van der Holst, Hermien Kan, **Erik Niks** (NETHERLANDS)

P11-07 | Adeno-Associated Virus Serotype 9 (AAV9) antibodies in patients with Spinal Muscular Atrophy Screened for treatment with Onasemnogene Abeparvovec

Eugenio Mercuri, John W. Day, Richard S. Finkel, Kathryn Swoboda, Elaine Kernbauer, Francis G. Ogrinc, Melissa Menier, Douglas M. Sproule, Douglas E. Feltner, Jerry R. Mendell (ITALY, USA)

P11-08 | Recurrent Hypokalemic Periodic Paralysis due to CACNA1S mutation

Marina Caner, Silvia Rial, Maribel Salguero, Belen Caurin, Veronica Perez, Diana Alvarez, Heidy Baide, **Gemma Olivé Cirera** (SPAIN)

P11-09 | Expanding the phenotype of Mitochondrial Thymidine Kinase 2 mutations

George Papadimas, Efthymia Vargami, Pinelopi Dragoumi, Rudy Van Coster, Joel Smet, Sarah Seneca, Constantinos Papadopoulos, Evangelia Kararizou, Dimitrios Zafeiriou (Greece, Belgium)

P11-10 | Evaluation of knowledge level of NUSINERSEN treatment in SMA families followed by a tertiary Paediatric Neurology clinic

Gulten Thomas, Olcay Unver, Eryim Karadag, Yasemin Gokdemir, Ela Erdem Eralp, Pinar Ergenekon, Ayca Evkaya, Sena Imamoglu, Adnan Dagcinar, Gunes Sager, Gulcan Akyuz, Hakki Akbeyaz, Bulent Karadag, Dilsad Turkdogan (TURKEY)

P11-11 | VIPN (Vincristine Induced Peripheral Neuropathy) – Neurological complications of Oncology treatment

Jadranka Sekelj Fureš, Sanja Pejić Roško, Lana Lončar, Vlasta Đuranović, Ivana Đaković, Katarina Vulin (CROATIA)

P11-12 | A paediatric case of anti-MuSK Antibody-Positive Ocular Myasthenia Gravis

Jeesuk Yu, Jeongju Hwang (REPUBLIC OF KOREA)

P11-13 | Clinical, genetic and neuropathological heterogeneity in a paediatric cohort with Nemaline Myopathy

Joana Martins, Jorge Oliveira, Cristina Garrido, Ricardo Taipa, Manuel Melo Pires, Manuela Santos (PORTUGAL)

Moderated Poster Track 3 (Sessions 8-11)

P11-14 | Nusinersen: A tertiary centre experience in Type 1 Spinal Muscular Atrophy

Joana Ribeiro, Andreia Lomba, Henrique Araújo, Vera Ribeiro, Núria Madureira, Filipe Palavra, Isabel Fineza (PORTUGAL)

P11-15 | Genetic testing for Indonesian Duchenne and Becker Muscular Dystrophy patients: the era of personalized medicine

Kristy Iskandar, Ery Dwianingsih, Sunartini, Poh San Lai (INDONESIA, SINGAPORE)

P11-16 | A newly defined Fukutin gene mutation in a child with Fukuyama-Type Congenital Muscular Dystrophy

Gurkan Bozan, **Kursat Bora Carman**, Eylem Kiral, Coskun Yarar, Hasan Bora Ulukapi, Bahattin Erdogan, Ener Cagri Dinleyici (TURKEY)

P11-17 | One year of Nusinersen treatment in Spinal Muscular Atrophy (SMA) in Hungary

Léna Szabó, Anita Gergely, Rita Jakus, Dorottya Czövek, Ágnes Herczegfalvi (HUNGARY)

P11-18 | The RESTORE Registry: a resource for measuring and improving Spinal Muscular Atrophy (SMA) outcomes

Laurent Servais, John W. Day, Darryl C. De Vivo, Janbernd Kirschner, Eugenio Mercuri, Francesco Muntoni, Perry B. Shieh, Eduardo Tizzano, **Marcus Droege**, Omar Dabbous, Farid Khan, Frederick A. Anderson, Richard S. Finkel (FRANCE, USA, GERMANY, ITALY, UNITED KINGDOM, SPAIN)

P11-19 | Burden of illness of Spinal Muscular Atrophy Type 1 (SMA1)

Marcus Droege, Omar Dabbous, Ramesh Arjunji, Jennifer Seda, Marjolaine Gauthier-Loiselle, Martin Cloutier, Douglas M. Sproule (USA, CANADA)

P11-20 | Paediatric Anti-3-Hydroxy-3-Methylglutaryl-Coenzyme A Reductase antibody associated Immune Necrotising Myopathy (anti-HMGCR INM) may mimic Limb Girdle Muscular Dystrophy in presentation

Marietta Pal-Magdics, **Mary Chesshyre**, Rahul Phadke, Mark Wood, Anna Sarkozy, Pinki Munot (UNITED KINGDOM)

P11-21 | Phenotypic variability in two maternal cousins with Calpainopathy

Mihaela Roxana Loghen, Diana Anamaria Epure, Raluca Ioana Teleanu (ROMANIA)

P11-22 | Limb-Girdle Muscular Dystrophy Type 2O as a clinical manifestation of POMGNT1 gene mutation

Gultekin Kutluk, **Naz Kadem** (TURKEY)

P11-23 | Electrophysiological diagnostic of neuromuscular diseases in new-borns, infants and toddlers

Philip Broser (SWITZERLAND)

P11-24 | Spinal rigidity, scoliosis and progressive respiratory impairment: clues to SEPN1 related Myopathy

Pinelopi Dragoumi, Euthymia Vargiami, Maria Kyriazi, Maria Milioudi, Dimos Gidaris, Fotios Kirvassilis, Dimitrios Zafeiriou (GREECE)

P11-25 | Muscle ultrasound comparison between early, intermediate and late onset Friedreich's Ataxia

Renate Verbeek, Anne Waalkens, Marieke Kuiper, Corien Verschuur-en-Bemelmans, Han van der Hoeven, Jeroen de Vries, Judith van Gaalen, Michel Willemsen, Berry Kremer, Katrin Bürk, **Deborah Sival** (NETHERLANDS, GERMANY)

P11-26 | Guillain-Barré Syndrome: analysis of Acute Inflammatory Neuropathies from a tertiary centre

Rita Martins, **Tiago Proença dos Santos**, Joana Coelho, Sofia Quintas, António Levy, Teresa Moreno (PORTUGAL)

P11-27 | Reservoir with a thoracic spinal catheter used for intrathecal delivery of Nusinersen in a patient with Type 2 Spinal Muscular Atrophy

Rocio Calvo-Medina, Sara Iglesias Moroño, Bienvenido Ros Lopez, Jose Miguel Ramos Fernandez, Maria Dolores Mora Ramirez (SPAIN)

Moderated Poster Track 3 (Sessions 8-11)

P11-28 | Neuromuscular Diseases in paediatric palliative care: a clinical challenge

Rocio Calvo-Medina, Ana Cabrera del Moral, Aurora Madrid Rodriguez, Maria Jose Pelaez Cantero, Miguel Angel Lendínez Ramirez, Tamara Fernandez Fernandez, Jose Miguel Ramos Fernandez (SPAIN)

P11-29 | Mortality and sedation at the end of the life in neurological patients in a paediatric palliative care unit

Rocio Calvo-Medina, Aurora Madrid Rodriguez, Maria Jose Pelaez Cantero, Jose Miguel Ramos Fernandez, Maria Dolores Mora Ramirez (SPAIN)

P11-30 | Intrathecal administration of Onasemnogene Abeparvovec gene-replacement therapy for Spinal Muscular Atrophy Type 2 (STRONG)

Richard S. Finkel, John W. Day, Basil T. Darras, Nancy L. Kuntz, Anne M. Connolly, Thomas O. Crawford, Russell J. Butterfield, Perry B. Shieh, Gihan Tennekoon, Susan T. Iannaccone, Matthew Meriggioli, Sitra Tauscher-Wisniewski, Francis G. Ogrinc, Sarah Kavanagh, Elaine Kernbauer, Joann Whittle, Douglas M. Sproule, Arseniy Lavrov, Douglas E. Feltner, Jerry R. Mendell (USA)

P11-31 | Dynein Heavy Chain Mutation: a case series

Samyami Chowdhury, Sithara Ramdas, Martin Smith, Sandeep Jayawant (UNITED KINGDOM)

P11-32 | Charcot-Marie-Tooth Disease associated with elevated creatine kinase and proteinuria due to Inverted Formin 2 (INF2) gene mutation

Ilknur Erol, Özgür Küük, Şeyda Beşen, Suzan Zorludemir, Aytül Noyan (TURKEY)

P11-33 | Early-onset Distal Myopathy related to a novel homozygous mutation in the MYH7 gene in a 17-year-old girl

Stephanie Schüssler, Katja Steinbrücker, Angela Abicht, Regina Trollmann (GERMANY)

P11-34 | Prevalence and genetic subtypes of Congenital Myasthenic Syndromes in Slovenian children

Anja Troha Gergeli, Tanja Golli, David Neubauer, Tita Butenko, Tanja Loboda, Damjan Osredkar (SLOVENIA)

P11-35 | Follow-up of 13 Duchenne boys treated with Ataluren in Portugal

Teresa Moreno, Manuela Santos, Cristina Garrido, Isabel Fineza, Joana Ribeiro (PORTUGAL)

P11-36 | Congenital Myopathies: clinicopathological findings and genotype-phenotype correlations

Yagmur Bayındır, Haluk Topaloğlu (TURKEY)

P11-37 | Intrathecal Nursinersen therapy in children with Spinal Muscular Atrophy. Our experience

Rocio Calvo-Medina, Esther Moreno-Medinilla, Maria Angeles Aviles Tirado, Pilar Diez del Corral Egea, Jose Miguel Ramos Fernandez, Maria Dolores Mora Ramirez (SPAIN)

P11-38 | Timed-function test data in patients with Duchenne Muscular Dystrophy from the STRIDE Registry and the CINRG Natural History Study: a matched cohort analysis

Eugenio Mercuri, Filippo Buccella, Isabelle Desguerre, Janbernd Kirschner, Francesco Muntoni, Andrés Nascimento Osorio, Már Tulinius, Abdallah Delage, Jin Zhu, Allan Kristensen, Panayiota Trifillis, Claudio L. Santos, Craig M. McDonald (ITALY, FRANCE, GERMANY, UNITED KINGDOM, SPAIN, SWEDEN, SWITZERLAND, USA)

P11-39 | Novel ANO5 homozygous mutation causing mild myalgia and unprovoked hyperkemia in an athlete teenager

Lais Carvalho, Allan Depizol, Ana Paula Bonatto, Zumira Carneiro, Charles Lourenco (BRAZIL)

P11-40 | Pontocerebellar Hypoplasia Type 1 (PCH Type 1): different phenotypes in patients with EXOSC3 mutations

Mireia Vazquez Piqueras, Begoña De Azúa, Patricia Fuentes Pita, Itxaso Martí, Verónica Delgadillo, Mónica Rebollo, Jordi Muchart, Belén Pérez Dueñas, Carmen Espinós, Delia Yubero, Judith Armstrong, Daniel Natera, Carlos Ortez, Andrés Nascimento, Angels García-Cazorla, Mar O'Callaghan, Alejandra Darling (SPAIN)

Moderated Poster Track 3 (Sessions 8-11)

P11-41 | The Identification of *CHRNE* 1267delG mutation in Greek Roma patients with Congenital Myasthenia
Chrysanthi Tsimakidi, Panagiota Athanasopoulou, Aikaterini Giannisi, Marina Katsalouli, Virginia Theodorou (GREECE)

P11-42 | The importance of the establishment a register for monitoring children with Neuromuscular Disorders
Tina Sabadin, Judita Kolenc (SLOVENIA)

P11-43 | Enlargement of peripheral nerves in late-onset Krabbe's Disease as demonstrated by nerve ultrasound
Hanna Küpper, Nadja Kaiser, Christiane Kehrer, Samuel Gröschel, Veronika Horber, Ute Grasshoff, Stefanie Beck-Wödl, Ingeborg Krägeloh-Mann, Alexander Grimm (GERMANY)

P11-44 | Extending the phenotype of the Hypotonia-Ataxia-Developmental Delay - Tooth Enamel Defect Syndrome (*CTBP1* gene)
Hanna Küpper, Joohyun Park, Andrea Bevot, Kathrin Grundmann-Hauser, Ute Grasshoff, Veronika Horber, Tobias Haack, Alexander Grimm, Ingeborg Krägeloh-Mann (GERMANY)

P11-45 | One-year follow-up of Slovenian DMD patients treated with Ataluren
Tanja Golli, Tita Butenko, Tanja Loboda, Damjan Osredkar (SLOVENIA)

P11-46 | From upper respiratory tract infection and myocarditis to the diagnosis of a child female carrier of Duchenne Muscular Dystrophy: a case report
Tita Butenko, Mirjana Perkovic Benedik, Sasa Bozicnik, Damjan Osredkar (SLOVENIA)

P11-47 | Evaluation of Slovenian children with Spinal Muscular Atrophy Type I-III six months after treatment with Nusinersen
Tita Butenko, Tanja Loboda, Tanja Golli, Damjan Osredkar (SLOVENIA)

P11-48 | Eculizumab as a long-term treatment in Congenital CD59 Deficiency: single-centre experience
Goknur Haliloglu, Nesibe Eroglu, Sule Unal, Mualla Cetin, Haluk Topaloglu (TURKEY)

P11-49 | Clinical course, outcome and autoantibody status in children with Chronic Demyelinating Polyneuropathy (CIDP)
Nina Barisic, Kevin Rostasy, Katerina Kiprianou, Ana Fluksek, Branka Bunoza, Kamelija Žarković, Desiree De Simoni, Romana Höftberger (CROATIA, GERMANY, AUSTRIA)

P11-50 | Genetic and nongenetic modifiers – Possible treatment targets in Spinal Muscular Atrophy
Nina Barisic, Ivan Lehman, Jadranka Sertić (CROATIA)

P11-51 | Phenotypic variability in siblings with Spinal Muscular Atrophy
Vedrana Milic Rasic, Vesna Brankovic, Ana Kosac, Slobodanka Todorovic, Zorica Stevic, Milos Brkusanin, Jovan Pesovic, Goran Brajuskovic, Dusanka Savic Pavicevic (SERBIA)

P11-52 | Dynamic Thiol/Disulphide homeostasis in children with Duchenne Muscular Dystrophy
Faruk Incecik, Gamze Avcioğlu, Özcan Erel, Salim Neşelioğlu, Ozlem M. Hergüner (TURKEY)

