Societatea Romana de Neurologie Pediatrica
Romanian Society of Pediatric Neurology

3rd National Conference of Romanian Society of Pediatric Neurology

October 26-28 2017

Crown Plaza Hotel, Bucharest

PRELIMINARY PROGRAMME

Thursday - October 26th

07.30 - 08.30 **Registration of participants**

08.30 - 08.40 **Opening of the Conference**

08.40 - 10.30 **Neurometabolic Disorders (part I):**
Chairs: Catrinel Iliescu, Linda de Meirleir

08.40 - 09.25 **Diagnosis algorithm for lysosomal disorders**
Linda de Meirleir, Brussels, Belgium

09.25 - 10.10 **Genetic testing for lysosomal disorders**
Alexander Gheldof, Brussels, Belgium

10.10 - 10.30 **Mucopolysaccharidoses – neurological aspects**
Mihaela-Adela Vintan, Camelia Al-Khzouz, Cătălin Dumitrache , Lori Mureșan Dumitrache - Cluj-Napoca, Romania

10.30 - 11.00 **Coffee break**

11.00 - 13.00 **Neurometabolic Disorders (part II)**
Chairs: Diana Bârcă,Carmen Sandu

11.00 - 11.50 **Congenital Glycosilation defects**
Linda de Meirleir, Brussels, Belgium

11.50 - 12.10 **Genetic testing in neurometabolic disorders – clinical case series**
Ursu Radu-Ioan, Cristina Dragomir, Bianca Basangiu, Gratiela Chelu, Oana Mantescu, Marinela Malangeanu, Sandra Grigore, Viorica Elena Radoi, Romania/ Synevo Diagnosis Center, Bucharest

12.10 - 12.30 **Glucose Transporter Type 1 Deficiency Syndrome (Glut1 DS) in pediatric movement disorders**
Diana Bârcă, Linda De Meirleir*, Nathalie Smeets*, Damien Lederer*, Carmen Burloiu
Bucharest, România/ *Brussels, Belgium

12.30 - 12.45 **Glutaric Acidemia Type 1 – clinical and evolution aspects based on a clinical case**
Mihaela-Adela Vintan, Carmen Culcițchi, Loria Mureșan, Cătălin Dumitrache
Cluj -Napoca, Romania

12.45 - 13.00 **Congenital cataract, a diagnostic clue in a rare disorder - Lowe syndrome**
Diana Bârcă, Linda De Meirleir*, Alexander Gheldof*, Oana Tarța-Arsene, Cristina Moțoescu
Bucharest România/*Brussels, Belgium

13.00 - 14.30 **Lunch and poster viewing.**

14.30 - 16.00 **Neurometabolic Disorders (part III)**
Chairs: *Voica Foișoreanu, Iuliu Bacoș Cosma*

14.30 - 14.50 **Diagnostic algorithm in leukodystrophies**
Diana Bârcă, Oana Tarța-Arsene, Catrinel Iliescu, Cristina Pomeran
Bucharest, Romania

14.50 - 15.05 **Muscle biopsy diagnostic value in neurometabolic diseases of childhood**
Alexandra Bastian, Emilia Manole, Colentina Clinical Hospital, Pathology Department, Bucharest, Romania

15.05- 15.20 **Genetic analyzes of neurometabolic disorders: a NGS approach**
Vasilica Plaiasu, Genetic Department, Mother and Child Institute, Bucharest, Romania

15.20 - 15.35 **Leber hereditary optic neuropathy - a cause of loss of visual acuity in teenagers**
Adrian Burloiu, Carmen Burloiu, Bucharest, Romania

15.35 - 15.50 **Wilson Disease: an update**
Andrei Marinescu, Alice Dică, Dana Craiu, Diana Bârcă
Bucharest, Romania

15.50 - 16.00 **CLN2 - where are we?**
Casandra Munteanu, Cristina Mincă, Diana Bârcă, Catrinel Iliescu
Bucharest, Romania

16.00 - 16.30 **Coffee break**

16.30 - 17.30 **Teaching session I - Pediatric multiple sclerosis (MS)(part I)**
(sustained by Merck Serono)
Chairs: Carmen Burloiu, Carmen Sandu

16.30 - 16.50 **Pediatric Multiple Sclerosis – cause and risk factors, the role of genetic and environmental factors**
Carmen Burloiu, Bucharest, Romania

16.50 - 17.10 **How we diagnose and assess children with MS**
Carmen Sandu, Bucharest, Romania

17.10 - 17.30 **Radiologically isolated syndrome - the relation with MS**
Carmen Burloiu, Bucharest, Romania

17.30 - 18.30 **Teaching session II - Pediatric multiple sclerosis (MS)**
(sustained by Johnson&Johnson)
Chairs: Dana Craiu, Cristina Pomeran, Alice Dica

17.30 - 17.50 **Clinical aspects of MS at pediatric age**
Alice Dica, Bucharest, Romania

17.50 - 18.10 **Clinical and outcome particularities in MS at small age**
Dana Craiu, Cristina Boghean, Carmen Burloiu,
Bucharest, Romania

18.10 - 18.30 **Current treatment concepts in pediatric MS**
Cristina Pomeran, Bucharest, Romania

18.30 - 19.30 **Teaching Session III - Actualities in Duchenne Muscular Dystrophy**
(sustained by Medison Pharma)
Chairs: Niculina Butoianu, Daniela Vasile

18.30 - 18.50 **Importance of genetic testing in DMD diagnostic**
Elena Neagu, Ligia Barbarii, National Institute of Legal Medicine ,
Bucharest, Romania

18.50 - 19.10 **Practical aspects regarding the management of patients with dystrophinopathies**
Daniela Vasile, "Victor Gomoiu"Hospital,
Bucharest, Romania

19.10 - 19.30 **Duchenne Muscular Dystrophy – innovative therapies**
Niculina Butoianu , Dana Surlica, Carmen Sandu, "Alex. Obregia Hospital"
Bucharest, Romania

19.30 - 21.30 **Opening Cocktail - Crown Plaza Hotel**

Friday – October 27th

08.30 - 10.30 **Epilepsy (part I) – Genetics Epilepsy in clinical practice**
Chairs : Dana Craiu, Cristina Moțoescu

08.30 - 08.50 **Epilepsy genetics – where we are in 2017?**
Dana Craiu, Bucharest, Romania

08.50 - 09.10 **The role of genetic testing in epilepsy management**
Catrinel Iliescu, Bucharest, Romania

09.10 - 09.30 **Genetic evaluation of the epileptic patients- clinical case series**
Viorica Elena Radoi, Cristina Dragomir, Bianca Basangiu, Gratiela Chelu, Oana Mantescu, Marinela Malangeanu, Sandra Grigore, Radu-Ioan,
Synevo Diagnosis Center, Bucharest, Romania

09.30 - 09.50 **Myoclono-atonic epilepsy – phenotypical variability starting from the classic phenotype**
Cristina Moțoescu, Bucharest, Romania

09.50 - 10.10 **Behind and beyond MECP2 gene**
Cristina Anghelescu, Dana Craiu*
LOTUS Medical Center, *Obregia Hospital, Bucharest, Romania

10.10 - 10.30 **Epilepsies in children with perinatal cerebral lesions - some diagnostic aspects**
Corina Grîu, Sprîncean Mariana, Calcâi Cornelia, Lupușor Nadejda, Revenco Nineli, Svetlana Hadjiu
Chișinău, Republique of Moldavia

10.30 - 11.00 **Coffee break**

11.00 - 12.30 **Epilepsy (part II) New epilepsy phenotypes.**
Chairs*: Ileana Benga, Oana Tarța-Arsene*

11.00 - 11.15 **Epilepsy associated with GRIN gene mutations.**
Oana Tarta-Arsene, Diana Bârcă, Corina Morar, Cristina Moțoescu,
Bucharest, Romania

11.15 - 11.30 **Refining the concept: Epileptic encephalopathies versus epilepsies with developmental encephalopathy – the role of the etiology.**
Dana Craiu, Bucharest, Romania

11.30 - 11.45 **Genetics of febrile seizures – what do we tell parents?**
Iuliu Bacoș-Cosma, Timișoara, Romania

11.45 - 12.00 **Epileptic spasms can have genetic causes**
Laura Popescu, Brașov, Romania

12.00 - 12.15 **SCN2A and familial neonatal seizures**
Carmen Sandu, Niculina Butoianu, Carmen Burloiu, Catrinel Iliescu,
Bucharest, Romania

12.15 - 12.30 **ESES: from clinics to EEG and back**
Cristina Pomeran, Carmen Sandu, Dana Craiu, Catrinel Iliescu, Diana Bârcă, Oana Tarța-Arsene, Cristina Moțoescu, Ioana Minciu, Alice Dica, Mădălina Leanca, Mădălina Lascu, Sanda Măgureanu,
Bucharest, Romania

12.30 - 13.00 **Epilepsy (part III) - Recommendations of medical practice and treatment of epilepsy**
Chairs: Catrinel Iliescu, Iuliu Bacoș-Cosma, Dana Craiu

12.30 - 12.45 **Recommendations of practice in Febrile Seizures.**
Dana Craiu

12.40 – 12.50 **Recommendations of practice in epileptic spasms/West Syndrome**
Catrinel Iliescu

12.50 – 13.00 **Recommendations of practice in Dravet Syndrome.**
Dana Craiu

13.00 - 14.30 **Lunch and poster viewing.**

14.30 - 16.00 **Cerebral malformations (part I)**
Chairs: Ioana Minciu, Anna Jansen, Alice Dica

14.30 - 14.50 **Cerebral malformations in children – generalities**
Diana Bârcă, Catrinel Iliescu, Bucharest, Romania

14.50 - 15.20 **The genetics of polymicrogiria revisited**
Anna Jansen, Brussels, Belgium

15.20 - 15.40 **Schizencephaly - what's behind?**
Alice Dica, Bucharest, Romania

15.40 - 16.00 **Clinical and etiological spectrum of lissencephaly**
Iuliu Bacoș Cosma, George Moisa,Timișoara, România

16.00 - 16.30 **Coffee break**

16.30 - 17.30 **Teaching Session IV – Tuberous Sclerosis (part I).**
Chairs: Dana Craiu, Anna Jansen

16.30 - 17.00 **Genetic syndromes with behavioural and cognitive symptomatology - the example of Tuberous Sclerosis Complex**
Anna Jansen, Brussels, Belgium

17.00 - 17.30 **mTOR pathway and Everolimus - what is the future** ?
Dana Craiu, Bucharest, Romania

17.30 - 18.30 **Teaching Session – Tuberous Sclerosis (part II).**
(sustained by Novartis)
Chairs: Iuliu Bacoș Cosma, Carmen Burloiu

17.30 - 17.50 **Everolimus - add-on treatment in pharmaco-resistant epilepsy in TSC – current recommendations**
Carmen Burloiu, Mădălina Leanca , Bucharest, Romania

17.50 - 18.05 **The anti-epileptic effect of Everolimus in patients with SEGA associated with TSC**
Madalina Leanca, Carmen Burloiu, Diana Barcă, Oana Tarța-Arsene, Catrinel Iliescu, Cristina Pomeran, Alice Dică, Carmen Sandu, Dana Craiu, Bucharest, Romania

18.05 - 18.20 **When we are thinking at epilepsy surgery in Tuberous Sclerosis- clinical case**
Oana Tarța -Arsene, Cristina Pomeran, Ruxandra Aursulesei, Ioana Mândruță, Sergiu Stoica, Alina Mitel, Carmen Burloiu, Bucharest, Romania

18.20 - 18.30 **Recommendations for clinical practice in TSC**
Mădălina Leanca, Iuliu Bacoș Cosma*, Carmen Burloiu
*Timişoara/București, România

18.30 - 19.30 RPNS General Assembly

SATURDAY – october 28th

09.00 - 10.30 Cerebral malformations (part II).

Chairs: *Svetlana Hadjiu, Gabriela Vișa, Magdalena Budișteanu*

9.00 - 9.20 **Investigation of the genetic mechanisms involved in brain malformations. The experience of the Department of Pediatric Neurology from " Prof.Dr. Alexandru Obregia" Clinical Hospital of Psychiatry**
Magdalena Budișteanu, Sorina Mihaela Papuc, Raluca Colesniuc, Ioana Borcan, Catrinel Iliescu Carmen Burloiu, Oana Tarta-Arsene, Diana Barca, Ioana Minciu, Bogdan Budisteanu, Ina Ofelia Focsa, Andreea- Cristina Tutulan-Cunita, Natalyia DiDonato, Dana Craiu, Aurora Arghir
Bucharest, Romania

9.20 - 9.40 **Holoprosencephaly – clinical and imagistic diagnosis, monitoring.**
Gabriela Adriana Vișa, Grațiela Ionescu, Livia Maria Ognean
Sibiu, Romania

9.40 - 10.00 **Focal cortical dysplasia in pediatric neurology patholgy**
Oana Tarța - Arsene, Dana Craiu, Diana Bârcă, Carmen Sandu, Carmen Burloiu, Catrinel Iliescu, Magdalena Budişteanu, Ioana Minciu, Cristina Pomeran, Cristina Moţoescu, Alice Dica, Mădălina Leanca, Sergiu Stoica, Anca Vişan, Adrian Iliescu, George Moisa, Ioana Mîndruţă, Niculina Butoianu,
Bucharest, Romania

10.00- 10.15 **Diagnostic aspects of stroke in newborns, infants and small children**
Mariana Sprincean , Nadejda Lupuşor, Corina Grîu, Cornelia Călcîi, Nineli Revenco, Setlana Hadjiu ,
Chisinău, Moldova Republique

10.15 - 10.30 **Reserved theme**

10.30 - 11.00 Coffee break

11.00 - 12.30 Cerebral malformations (part III).

Chairs: Ioana Minciu, Adrian Toma

11.00 - 11.30 **Structural disorderes of the cerebellum and brainstem. Classification and clinical cases**
Adrian Ioan Toma, Medlife Memorial Hospital, Bucharest, Romania

11.30 - 11.50 **Joubert Syndrome spectrum**
Catrinel Iliescu, Carmen Burloiu, Bucharest, Romania

11.50 - 12.10 **Pontocerebellar Hypoplasia Type 2- a rare cause of developmental disorder**
Carmen Sandu, Alina Mitel, Carmen Burloiu, Bucharest, Romania

12.10 - 12.30 **The role of the ultrasound in the diagnosis and monitoring of the abnormalities of the posterior fossa and spinal malformations**
Adrian Ioan Toma, Life Memorial Hospital, București, România

12.30 - 12.50 **Galen vein malformation**
Stefăniță Dima, Adrian Ioan Toma, Life Memorial Hospital, Bucharest, Romania

12.50 - 13.10 Presentation of awaerd winning posters

13.10 - 13.50 Highlights in Pediatric Neurology
Dana Craiu, Diana Bârcă, Catrinel Iliescu, Carmen Burloiu, Nina Butoianu, Ioana Minciu

13.50 - 14.00 Conclusions. Closure of the Congress