

50^{ème} Réunion
de la SENP+
50th SENP+
Meeting

Société Européenne
de Neurologie Pédiatrique

14, 15 & 16 mars 2024

Milan, Italie



COMITÉS / COMMITTEES

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Organizing local committee

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LIEU DU CONGRÈS / CONGRESS VENUE

Université de Milan / University of Milan
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SOMMAIRE / AGENDA

Edito / Editorial	p.4
Planning	p.6
Programme / Program	p.7
Jeudi 14 Mars / Thursday, March 14	p.7
Vendredi 15 Mars / Friday, March 15	p.8
Samedi 16 Mars / Saturday, March 16	p.12
Posters	p.15
Symposiums / Symposia	p.26
Sponsors	p.27
Social program / Programme social	p.28



EDITO

Chers collègues et amis,

Au nom de la Société européenne de neurologie pédiatrique et du comité scientifique de l'organisation locale, nous sommes heureux de vous accueillir à Milan pour la 50^e réunion de la SENP+.

La 50^e réunion de notre société représente une étape unique qui démontre que la SENP+ a des racines anciennes et solides et qu'aujourd'hui nous continuons sur les traces d'éminents chercheurs qui ont marqué l'histoire de la neurologie pédiatrique européenne. C'est un grand honneur, mais aussi une tâche dont nous espérons nous acquitter lors de cette réunion, qui se tiendra pour la première fois à Milan, dans les locaux de l'université qui célèbre son premier centenaire ces jours-ci.

Les deux thèmes choisis cette année seront :

- Les maladies héréditaires de la substance blanche cérébrale
- Les maladies infectieuses et para-infectieuses du SNC et du SNP

Les maladies héréditaires de la substance blanche sont un sujet qui a connu un énorme développement ces dernières années, grâce à la découverte de la génétique et au typage neuroradiologique, avec un cadrage de plus en plus précis. Les propositions thérapeutiques les plus récentes et la possibilité d'études collaboratives à réaliser au sein de la société seront également présentées.

Le second thème est d'une grande importance clinique en raison de la fréquence d'apparition de ces maladies, des questions qui restent en suspens, des protocoles d'intervention qui sont encore en cours d'amélioration, mais aussi parce que, dans

l'ère que nous vivons après la pandémie de Covid, de nouveaux types de troubles sont apparus que nous apprenons à mieux connaître et à mieux comprendre.

Ces thèmes seront abordés par des orateurs prestigieux de différents pays et, comme toujours, nous comptons sur la contribution active de nombreux jeunes qui sont la force vive de chaque congrès SENP+.

En raison du 50^e anniversaire, cette édition marquera un moment de réflexion pas seulement sur les sujets spécifiés ci-dessus, mais aussi sur comment la connaissance, l'interprétation et la thérapie des pathologies ont évolué au cours de ces 50 années et quelles contributions les membres de la SENP+ ont pu apporter à ce cheminement.

Dans le cadre des célébrations du 50^e anniversaire, un livre sur l'histoire de notre société sera présenté, ce qui permettra aux plus jeunes de connaître les bases sur lesquelles SENP+ a été fondée et les personnes qui ont contribué à la rendre grande.

Nous vous donnons donc rendez-vous à Milan, une ville vivante, accueillante et stimulante qui saura ajouter à la grande profondeur scientifique de la réunion un accueil chaleureux et familial, comme le veut la tradition des réunions de la SENP+.

Profitez tous de notre congrès à Milan !

Au nom du CO et du CS local,
Pierangelo Veggiotti



EDITORIAL

Behind us, a great history to build a brilliant future.

On behalf of the European Society of Pediatric Neurology and the scientific committee of the local organization, we are pleased to welcome you to Milan for the 50th meeting of the ESPN.

The 50th meeting of our organization represents a unique milestone that shows the deep and strong roots of the SENP, and today we continue in the footsteps of eminent researchers who have shaped the history of European pediatric neurology. It is a great honor but also a task that we hope to fulfill during this meeting, which will be held for the first time in Milan, at the premises of the university that is celebrating its centenary these days.

The two themes chosen this year are:

- Hereditary diseases of the cerebral white matter
- Infectious and para-infectious diseases of the CNS and PNS

Hereditary diseases of the white matter are a subject that has seen enormous development in recent years, thanks to the discovery of genetics and neuroradiological typing, with increasingly precise framing. The most recent therapeutic proposals and the possibility of collaborative studies to be carried out within the organization will also be presented.

The second theme is of great clinical importance due to the frequency of occurrence of these diseases, the unanswered questions, the ongoing improvement

of intervention protocols, but also because in the era we live, after the Covid pandemic, new types of disorders have emerged that we are learning to know and understand better.

These themes will be addressed by prestigious speakers from different countries, and as always, we count on the active contribution of many young researchers who are the driving force of every SENP congress.

Due to the 50th anniversary, this edition will be a moment of reflection not only on the topics mentioned above, but also on how the knowledge, interpretation, and therapy of pathologies have evolved over these 50 years and what contributions the members of the SENP have been able to make to this journey.

As part of the 50th-anniversary celebrations, a book on the history of our organization will be presented, which will allow younger individuals to learn about the foundations on which SENP was founded and the people who have contributed to its greatness.

We look forward to seeing you in Milan, a vibrant, welcoming, and stimulating city that will add a warm and familial atmosphere to the great scientific depth of the meeting, as is customary in SENP congresses.

Enjoy our congress in Milan !

Pierangelo Veggiotti

14 MARS / MARCH 14

COURS DÉDIÉ AUX PÉDIATRES / COURSE DEDICATED TO PEDIATRICIANS

Urgences en neurologie pédiatrique / Emergencies in child neurology

13h30	Accueil / Welcome
14h00	Conférences / Lectures
16h00	Conférences / Lectures
18h00	Présentation du Livre / Book Presentation
19h00	Cérémonie d'ouverture / Opening Ceremony
20h00	Cocktail de Bienvenue / Welcome Cocktail

15 MARS / MARCH 15

Les maladies héréditaires de la substance blanche cérébrale

Hereditary diseases of the cerebral white matter

07h30	Accueil / Welcome
08h00	Ouverture du congrès / Congress opening
08h30	Conférences / Lectures
10h00	Communications orales / Oral communications
11h00	Symposium sponsorisé par l'industrie / Industry sponsored symposium
13h00	Symposium sponsorisé par l'industrie / Industry sponsored symposium
14h00	Session Poster Flash / Flash Poster Session
14h30	Communications orales / Oral communications
16h00	Conférences / Lectures
17h30	Assemblée Générale de la SENP / SENP General Assembly
19h30	Visite guidée du Musée des sciences et des techniques Léonard de Vinci Guided visit the Leonardo da Vinci Museum of Science and Technology (pour les participants au dîner de gala uniquement / for gala dinner participants only)
20h30	Dîner de gala / gala dinner

16 MARS / MARCH 16

Pediatric Pediatric neuroinflammatory disorders of infectious etiology

Infectious and para-infectious diseases of the CNS and PNS

08h00	Visite des Posters / Poster Visit
08h30	Conférences / Lectures
10h30	Conférences / Lectures
12h00	Visite Poster / Poster Visit
14h00	Discussion de Cas Cliniques / Clinical Cases Discussion
15h00	Communications orales / Oral communications
16h00	Remise prix & clôture du congrès / Awards Ceremony & Farewell



PROGRAMME / PROGRAM

JEUDI 14 MARS
THURSDAY, MARCH 14

COURS DÉDIÉ AUX PÉDIATRES / COURSE DEDICATED TO PEDIATRICIANS

Urgences en neurologie pédiatrique / Emergencies in child neurology

13h30 Accueil / Welcome

Conférences / Lectures

Modérateurs / Chairs : Gianvincenzo Zuccotti, Valentina Fabiano, Stefania Bova

14h00 Migraïne and headache / Migraïne et céphalée
Caroline Ménache - Hirslanden Clinique des Grangettes, Geneva, Switzerland

14h30 Prise en charge d'une 1ère crise épileptique chez l'enfant / Management of a first epileptic seizure in children
Ilhem Ben Youssef Turki - Université Tunis El Manar, Tunis, Tunisia

15h00 Status epilepticus and recurrent seizures/ Statut épileptique et crises récurrentes
Alec Aeby - Université libre de Bruxelles, Brussels, Belgium

15h30 Pause-café / Coffee break

Conférences / Lectures

Modérateurs / Chairs : Gianvincenzo Zuccotti, Valentina Fabiano, Stefania Bova

16h00 Ataxies aiguës / Acute ataxias
François Rivier - Université de Montpellier, Montpellier, France

16h30 Pediatric ischemic stroke: from initial symptoms to clinical management / Accident vasculaire cérébral ischémique pédiatrique : des premiers symptômes à la prise en charge clinique
Federica Teutonico - Ospedale Niguarda Ca' Granda, Milan, Italy

17h00 Functional Neurological disorders / Troubles neurologiques fonctionnels
Nardo Nardocci - Istituto Neurologico C Besta, Milan, Italy

18h00 Présentation du Livre / Book Presentation

19h00 Cérémonie d'ouverture avec Marcello Massimini / Opening Ceremony with Marcello Massimini

20h00 Cocktail de Bienvenue / Welcome Cocktail



PROGRAMME / PROGRAM

VENDREDI 15 MARS
FRIDAY, MARCH 15

Les maladies héréditaires de la substance blanche cérébrale / Hereditary diseases of the cerebral white matter

07h30 Accueil / Welcome

08h00 Ouverture du congrès / Congress opening

Conférences / Lectures

Modérateurs / Chairs : Davide Tonduti, Elisa Fazzi

- 08h30 The history of leukodystrophies: from disease definition to understanding and therapy / L'histoire des leucodystrophies : de la définition de la maladie à la compréhension et à la thérapie
Marjo van der Knaap - Amsterdam University Medical Centers, Amsterdam, The Netherlands
- 09h00 Understanding leukodystrophies at its core: the vital contribution of neuropathology / Comprendre les leucodystrophies à leur essence : la contribution vitale de la neuropathologie
Marianna Bugiani - Amsterdam University Medical Centers, Amsterdam, The Netherlands
- 09h30 From discovery to hope: a journey through the history of metachromatic leukodystrophy / De la découverte à l'espoir : un voyage à travers l'histoire de la leucodystrophie métachromatique
Francesca Fumagalli - San Raffaele Hospital, Milan, Italy

Communications orales / Oral communications

Modérateurs / Chairs : Joel Fluss, Jean Paul Misson

- 10h00 **CO01** – Clinical and genetic study of leukodystrophies in Tunisian cohort
Ichraf Kraoua - Lr18sp04 and Department of Pediatric Neurology, National Institute Mongi Ben Hmida of Neurology of Tunis, Tunisia
- 10h12 **CO02** – Preliminary data from the analysis of neuroradiological findings in Type I Alexander Disease
Ylenia Vaia - Unit of Pediatric Neurology, C.o.a.l.a. (center For Diagnosis and Treatment of Leukodystrophies), V. Buzzi Children's Hospital, Università Degli Studi Di Milano, Italy
- 10h24 **CO03** – Multicenter International Cohort of p.Ala177Thr AGS2 homozygous mutated patients: clinical features, disease evolution and search for prognostic features
Costanza Varesio - Department of Brain And Behavioral Sciences, University Of Pavia, Department Of Child Neurology And Psychiatry, Irccs Mondino Foundation, Italy



- 10h36 **CO04** – Clinical investigation coupled with molecular dissection as the personalized approach to pediatric GNAO1 encephalopathies. Drug discovery and the potential to other rare diseases
Vladimir Katanaev – University of Geneva, Geneva, Switzerland
- 10h48 **CO05** – Presentation Modes in Alexander Disease: A Multicenter Retrospective Study of 94 Patients
Emma Perrier – Service De Neuropédiatrie, Cmr Neurogénétique, Hôpital Armand Trousseau-La Roche Guyon, Aphp, Sorbonne University, Paris, France
- 11h00 Symposium sponsorisé par l'industrie / Industry sponsored symposium
(See page 13 for the detailed program)
- 12h00 Déjeuner et Visite des Posters / Lunch & Poster Visit
- 13h00 Symposium sponsorisé par l'industrie / Industry sponsored symposium
(See page 13 for the detailed program)
- Session Poster Flash / Flash Poster Session
Modérateurs/Chairs : Ilhem Ben Youssef Turki, Concita Escofet Soterias
- 14h00 **PF01** – Acute necrotizing encephalopathy associated with RANBP2 mutation: case presentation and literature review
Irene Peterlongo – Department of Biomedical And Clinical Sciences, Postgraduate School Of Child Neuropsychiatry, University Of Milan, Italy
- 14h06 **PF02** – Hypomyelinating leukodystrophies associated with GJC2 gene mutations: a Tunisian case series
Ichraf Kraoua – Lr18sp04 and Department of Pediatric Neurology, National Institute Mongi Ben Hmida of Neurology of Tunis, Tunisia
- 14h12 **PF03** – The first Italian single center cohort of Phelan McDermid syndrome patients: clinical and genetic aspects
Alice Decio – Irccs Medea, Italy
- 14h18 **PF04** – Neurodevelopmental profile in children born to mothers affected by Systemic Sclerosis
Erika Loi – Department of Clinical and Experimental Sciences, University of Brescia; Unit Of Child Neurology And Psychiatry, Asst Spedali Civili Of Brescia, Italy
- 14h24 **PF05** – Cognitive skills in Aicardi Goutières Syndrome
Annamaria Del Boca – Department of Brain and Behavioral Sciences, University of Pavia, Department of Child Neurology and Psychiatry, Irccs Mondino Foundation, Italy



PROGRAMME / PROGRAM

VENDREDI 15 MARS
FRIDAY, MARCH 15

Communications orales / Oral communications

Modérateurs / Chairs : Ichraf Kraoua, Jessica Galli

- 14h30 **CO06** – Clinical and genetic characterization of PLP1-related disorders: preliminary insights from an Italian cohort
Fabio Bruschi – Pediatric Neurology Unit, Coala (center For Diagnosis and Treatment of Leukodystrophies) V. Buzzi Hospital, Università Degli Studi di Milano, Italy
- 14h42 **CO07** – Stroke and stroke-like episodes as a new clinical manifestation in GLUT1 Deficiency Syndrome
Roberto Previtali – University of Milan, Italy
- 14h54 **CO08** – Study of children with neurological, metabolic and/or neurosurgical pathology transferred from Antilles Guyane to a Parisian university hospital from January 1, 2018 to January 1, 2023
Léa Lacoffrette – Department of Pediatrics, Martinique University Hospital, University of The French West Indies and French Guiana, France
- 15h06 **CO09** – Hidden Challenges: Mental Health Outcomes in Pediatric Stroke Survivors
Ludovica Serafini – Neurology, The Hospital for Sick Children, Toronto, Canada
- 15h18 **CO10** – Developmental coordination disorder and Cerebral visual impairment: convergences and divergences
Serena Micheletti – Unit of Child Neurology and Psychiatry, Asst Spedali Civili of Brescia, Italy
- 15h30 Pause-café et Visite des Posters / Coffee Break and Poster Visit

Conférences / Lectures

Modérateurs / Chairs : Simona Orcesi, Monica Vasconcelos

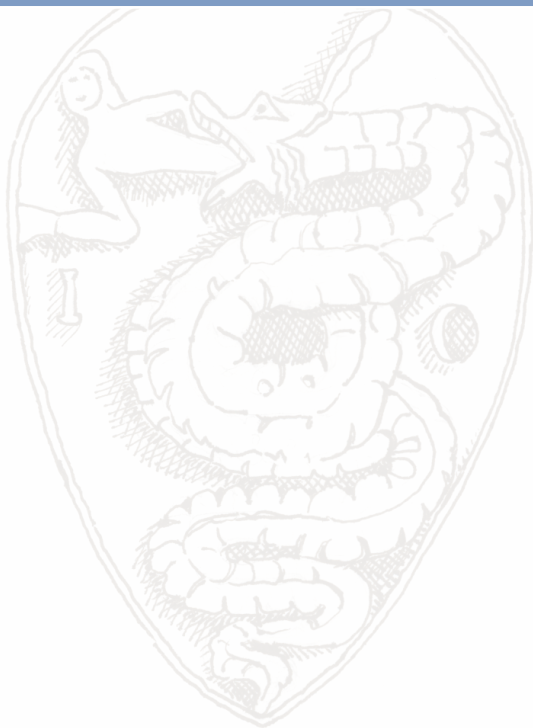
- 16h00 Pelizaeus-Merzbacher Disease and “PLPathies” from clinic to therapies / La maladie de Pelizaeus-Merzbacher et les “PLPathies” : de la clinique aux thérapies
Odile Boespflug Tanguy – Neurodiderot Université de Paris Cité, Paris, France
- 16h30 Challenges and advances in managing Aicardi-Goutières Syndrome / Les défis et les avancées dans la prise en charge du syndrome d'Aicardi-Goutières”
Elisa Fazzi – Università degli Studi di Brescia, Brescia, Italy
- 17h00 X-Linked Adrenoleukodystrophy: Past, Present, and Future Directions / Défis et avancées dans la prise en charge du syndrome d'Aicardi-Goutières
Caroline Sevin – Hôpital du Kremlin Bicêtre, Paris, France



17h30 Assemblée Générale de la SENP/ SENP General Assembly

19h30 **Visite guidée du Musée des sciences et des techniques Léonard de Vinci**
Guided visit the Leonardo da Vinci Museum of Science and Technology
(pour les participants au dîner de gala uniquement / for gala dinner participants only)

20h30 Dîner de gala / gala dinner





PROGRAMME / PROGRAM

SAMEDI 16 MARS
SATURDAY, MARCH 16

*Les maladies héréditaires de la substance blanche cérébrale / Pediatric
neuroinflammatory disorders of infectious etiology*

08h00 Visite Poster / Poster visit

Conférences / Lectures

Modérateurs / Chairs : Filipe Palavra, Jaime Campistol

- 08h30 Evolution and changing paradigms of diagnostic perspectives and patient management in pediatric infectious and post-infectious encephalitis / Évolution et changement de paradigmes dans les perspectives diagnostiques et la prise en charge des patients atteints d'encéphalite infectieuse et post-infectieuse pédiatrique
Yael Hachohen – University College London, London, United Kingdom
- 09h00 From traditional pathology to modern challenges: the evolution of neuropathology in the field of infectious and parainfectious disorders of CNS / De la pathologie traditionnelle aux défis modernes : l'évolution de la neuropathologie dans le domaine des troubles infectieux et parainfectieux du système nerveux central
Marianna Bugiani – Amsterdam University Medical Centers, Amsterdam, The Netherlands
- 09h30 Neuroradiological Aspects of Infectious Encephalitis: from the Typical Phenotype to the Challenge of Aspecific findings / Aspects neuroradiologiques de l'encéphalite infectieuse : du phénotype typique au défi des résultats aspécifiques
Cecilia Parazzini – Ospedale dei Bambini Buzzi, Milan, Italy
- 10h00 Pause-café et Visite des Posters / Coffee Break and Poster Visit
- 10h30 Human genetic and immunological determinants of childhood HSV1 encephalitis: a 20-year long journey / Déterminants génétiques et immunologiques humains de l'encéphalite à HSV1 chez l'enfant : un parcours de 20 ans
Shen-Ying Zhang – The Rockefeller University, New York, United States
- 11h00 Pleomorphic neurological acute presentations of pediatric Lyme LNB and Varicella-Zoster virus: lessons from the past and on-going controversies / Présentations neurologiques aiguës polymorphes de la maladie de Lyme (LNB) et du virus de la varicelle-zona chez les enfants : enseignements du passé et controverses actuelles
Joël Fluss – Hôpitaux Universitaires Genève, Geneva, Switzerland



- 11h30 Acute flaccid myelitis: emerging polio-like disease in an era believed to be polio-free? / Myélite flasque aiguë : maladie émergente de type polio dans une ère considérée comme exempte de polio
Jelte Helfferich - University Medical Center Groningen, Groningen, The Netherland

12h00 Visite des posters / Poster visit

12h30 Déjeuner et Visite des Posters / Lunch & Poster Visit

Discussion de Cas Cliniques / Clinical Cases Discussion
Modérateurs/Chairs : Nardo Nardocci, Roberta Cilio

- 14h00 **DDC1** - Epilepsy surgery, ketogenic diet or acetazolamide in a boy with refractory epilepsy
Benoît Semal - Université Libre De Bruxelles, Hôpital Universitaire De Bruxelles, Hôpital Universitaire Des Enfants Reine Fabiola, Department Of Genetics, Belgium

- 14h15 **DDC2** - Severe Inflammatory lesions with partial Kluver-Bucy Syndrome. New infectious or Autoimmune phenotype?
Ana Roche - Hospital Parc Taulí De Sabadell / Hospital Clínic De Barcelona, Spain

- 14h30 **DDC3** - A deceiving diagnostic process in an infant with acute encephalopathy. When clinical findings, EEG and neuroimaging disagree: which path to follow?
Ylenia Vaia - Unit of Pediatric Neurology, C.o.a.l.a. (center For Diagnosis and Treatment of Leukodystrophies), V. Buzzi Children's Hospital, Università Degli Studi di Milano, Italy

- 14h45 **DDC4** - Subacute onset of encephalopathy with movement disorder: diagnostic challenges between old and new diagnosis
Caterina Zanus - Institute for Maternal and Child Health Burlo Garofolo, Italy

Communications orales / Oral communications
Modérateurs / Chairs : Maria Paola Canevini, Irene Bagnasco

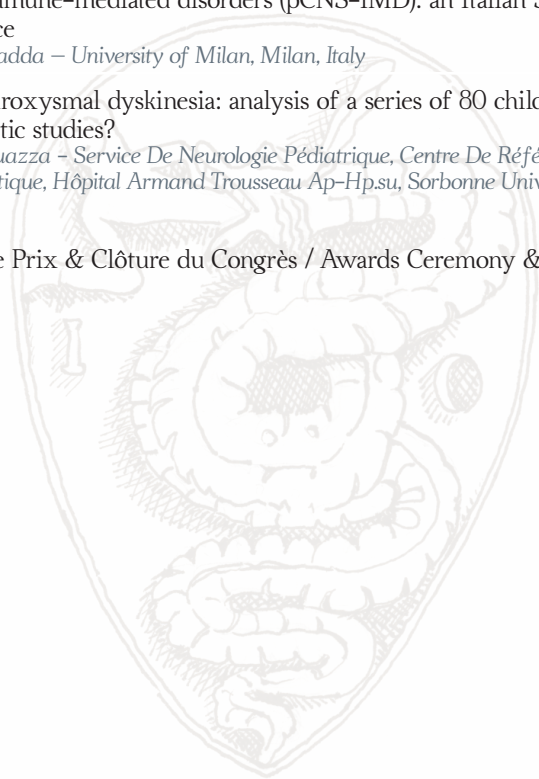
- 15h00 **CO11** - Children with Autism Spectrum Disorder can imagine actions like their neurotypical peers?
Jessica Galli - Department of Clinical And Experimental Sciences, University Of Brescia And Unit Of Child Neurology Psychiatry, Asst Spedali Civili Of Brescia, Brescia, Italy
- 15h12 **CO12** - Electroclinical phenotype and possible genotype-phenotype correlations in Pallister Killian syndrome: preliminary data
Giulia Ferrera - University Of Milan, Italy
- 15h24 **CO13** - Early discontinuation of treatment in neonates with acute provoked seizures: a European, multicenter, retrospective study
Margherita Bonino - Cliniques Saint-Luc U.C Louvain, Belgium



RELAZIONI & CORSO

SAMEDI 16 MARS
SATURDAY, MARCH 16

- 15h36 **CO 14** – Toward a phenotype characterization at onset in pediatric central nervous system immune-mediated disorders (pCNS-IMD): an Italian Single-Center Experience
Arianna Gadda – University of Milan, Milan, Italy
- 15h48 **CO15** – Paroxysmal dyskinesia: analysis of a series of 80 children, back to the basis after genetic studies?
Floriane Quazza – Service De Neurologie Pédiatrique, Centre De Référence De Neurogénétique, Hôpital Armand Trousseau Ap-Hp.su, Sorbonne Université, Fhu I2-D2, France
- 16h00 Remise de Prix & Clôture du Congrès / Awards Ceremony & Farewell





POSTERS

- P01 About use of N-acetyl -l-leucine (NALL) in a juvenile form of Tay Sachs Disease
Margarita CASTRO REY, Antonio Jesus MORALES MORENO, Laura MORALES ALBERTOS, Selma VAZQUEZ MARTIN, Carmen ALONSO VICENTE, Jose Manuel MARUGAN DE MIGUELSANZ, Jair ALONSO FERRERO - (1)Pediatrics, Spain
- P02 Ketogenic diet in developmental and epileptic encephalopathies
Margarita CASTRO REY, Selma VAZQUEZ MARTIN, Carmen ALONSO VICENTE, Laura MORALES ALBERTOS, Maria DE FELIPE PEREZ, Antonio Jesus MORALES MORENO, Jair ALONSO FERRERO, Jose Manuel MARUGAN DE MIGUELSANZ - (1)Pediatrics, Spain
- P03 Mathematical cognition: facts, difficulties and impacts on school learning
Marie-Jose PENNIELLO-VALETTE (1), Delphine CLERVAL (2), Hélène DESMAISONS (2), Séverine FRABOULET (3), Catherine MULLER MOULIN (4), Vanessa PAIN (4), Christelle BOLLORE (4) - (1)Neurologue Crtla Chu De Caen, France, (2)Orthophoniste Crtla Chu De Caen, France, (3)Neuropsychologue Crtla Chu De Caen, France, (4)Orthophoniste, France
- P04 MERS (Mild encephalopathy with reversible splenic lesion): case reports of two different clinical presentations
Alice POMPILI (1), Linda GASPARINI (2), Stefania BERGAMONI (2), Caroline REGNA-GLADIN (2), Paola DONEDA (2), Aglaia VIGNOLI (3) - (1)Università Degli Studi Di Milano, Italy, (2)Asst Grande Ospedale Metropolitano Di Niguarda, Italy, (3)Asst Grande Ospedale Metropolitano Di Niguarda-Università Degli Studi Di Milano, Italy
- P05 Equine Assisted Therapy in a sample of children with Autism Spectrum Disorder
Alessia LEIDI (1), Alice PASSARINI (1), Aurora SOTGIU (1), Michela RICEPUTI (1), Annalisa MARNONI (1), Alessia CARUSO (1), Aglaia VIGNOLI (2) - (1)Grande Ospedale Metropolitano Niguarda, Italy, (2)Grande Ospedale Metropolitano Niguarda-Università Degli Studi Di Milano, Italy
- P06 DDX3X syndrome: A common cause of intellectual disability in women
Gemma AZNAR-LAÍN (1), Gabriela Claudia SECONDI (2), Luis Alberto PÉREZ-JURADO (3) (1)pediatric Neurology Hospital Del Mar, Spain, (2)Pediatric Neurology Hospital Del Mar, Espagne, (3)Genetics Service. Hospital Del Mar Research Institute (imim), Spain
- P07 Impact of stiripentol on seizure-free days and new open label data from the STICLO clinical trials confirm its significant impact on seizure reduction and on quality of life in Dravet syndrome
Renzo GUERRINI (1), Perrine HUGON (2), Benjamin SERRAZ (2), Laurent CHANCHARME (3), Catherine CHIRON (4) - (1)Pediatric Neurology, Neurogenetics And Neurobiology Unit And Laboratories, Neuroscience Department, A. Meyer Children's Hospital, University Of Florence, Italie, (2)Biocodex, Orphan Disease Division (hq), France, (3)Biocodex, R&d Department, France, (4)Inserm U1141 & Aphp, Necker-Enfants Malades Hospital, Pediatric Neurology And Reference Center For Rare Epilepsies, France



POSTERS

- P08 Comparative efficacy and safety of stiripentol, cannabidiol and fenfluramine as first-line add-on therapies for the treatment of seizures in Dravet syndrome (DS): a network meta-analysis study
Renzo GUERRINI (1), Catherine CHIRON (2), Perrine HUGON (3), Delphine VANDAME (3), Warren LINLEY (4), Toby TOWARD (5) – (1)Pediatric Neurology, Neurogenetics And Neurobiology Unit And Laboratories, Neuroscience Department, A. Meyer Children's Hospital, University Of Florence, Italy, (2)Inserm U1141 And Aphp, Necker-Enfants Malades Hospital, Pediatric Neurology, France, (3)Biocodex, Orphan Disease Division (hq), France, (4)Paragon Market Access Ltd, Chorley, United Kingdom, (5)Henley Health Economics Ltd, United Kingdom
- P09 Ketogenic diet in epilepsy in the first year of life: indications and management
Roberto PREVITALI (1), Ilaria SERATI (1), Ramona DE AMICIS (2), Sara OLIVOTTO (3), Stefania Maria BOVA (3), Simona BERTOLI (2), Pierangelo VEGGIOTTI (3) – (1)University Of Milan, Italy, (2)Icans-Dis, Department Of Food Environmental And Nutritional Sciences, Italy, (3)Pediatric Neurology Unit, Buzzi Children's Hospital, Italy
- P10 Acute neurological manifestations of infectious conditions correlated with respiratory syncytial virus: an underestimated phenomenon? Difficulties in real-life patient management
Nicoletta ZERBI (1), Emilia RICCI (1), Ilaria VIGANO (2), Valentina CHIESA (2), Francesca LA BRIOLA (2), Samuele PALAZZO (2), Costanza PARAMITHIOTTI (2), Elisabetta SALVATICI (2), Giuseppe BANDERALI (1), Maria Paola CANEVINI (1) – (1) Unimi, Italy, (2) Asst Santi Paolo E Carlo, Italy
- P11 ACOX1 deficiency: early diagnosis through newborn screening program for X-linked adrenoleukodystrophy
Eleonora BONAVENTURA (1), Luisella ALBERTI (2), Andrea META (2), Maria IASCONI (3), Ilaria BOSIO (4), Laura MALERBA (5), Chiara MONTANARI (6), Cecilia PARAZZINI (7), Luigina SPACCINI (8), Cristina CEREDA (2), Davide TONDUTI (1) – (1)Child Neurology Unit, V. Buzzi Children's Hospital, Italy, (2)Newborn Screening And Inherited Metabolic Disease Unit, V. Buzzi Children's Hospital, Italy, (3)Molecular Genetics Laboratory, Asst Papa Giovanni Xxiii, Italy, (4)Department Of Neonatology And Neonatal Intensive Care Unit, Children Hospital, Asst Spedali Civili, Italy, (5)Unit Of Child Neurology And Psychiatry, Asst Spedali Civili, Italy, (6)Department Of Paediatrics, V. Buzzi Children's Hospital, Italy, (7)Paediatric Radiology And Neuroradiology Department, V. Buzzi Children's Hospital, Italy, (8)Clinical Genetics Unit, V. Buzzi Children's Hospital, Italy
- P12 The expanding knowledge of epilepsy in leukodystrophies
Eleonora MINACAPILLI (1), Ylenia VAIA (1), Silvia MASNADA (2), Pierangelo VEGGIOTTI (3), Davide TONDUTI (4), Monica Anna Maria LODI (2) – (1)Department Of Biomedical And Clinical Sciences, Postgraduate School Of Child Neuropsychiatry, University Of Milan, Italy, (2)Pediatric Neurology Unit, Buzzi Children's Hospital, Italy, (3)Pediatric Neurology Unit, Buzzi Children's Hospital; Department Of Biomedical And Clinical Sciences, University Of Milan, Italy, (4)Pediatric Neurology Unit, Buzzi Children's Hospital; C.o.a.l.a. (center For Diagnosis And Treatment Of Leukodystrophies), Buzzi Children's Hospital, Italy



POSTERS

- P13 The contribution of Epidyolex to the treatment of developmental and epileptic encephalopathies
Margarita CASTRO REY, Mario URBANO MARTIN, Selma VÁZQUEZ MARTIN, Ramon CANCHO CANDELA, Virginia NAVARRO ABIA, Aranzazu HERNANDEZ FABIAN, Eva Maria DOMINGUEZ BERNAL, Aquilina JIMENEZ GONZALEZ, Sandra TERROBA SEARA, Cristina RODRIGUEZ FERNANDEZ - (1)Pediatrics, Spain
- P14 Focal dystonia associated with Grisel Syndrome
Margarita CASTRO REY, Jair ALONSO FERRER, Laura ESCOBAR FERNANDEZ, Rosalia ELICES CRESPO, Elena URBANEJA RODRIGUEZ, Fernando CENTENO MALFAZ, Albertos MORALES, Antonio MORENO - (1)Pediatrics, Spain
- P15 Microdeletion 15q13.3: clinical spectrum with epileptic encephalopathy and customized treatment
Ana ROCHE, Débora ITZEP, Marta MORALEDA, Conxita ESCOFET, María Jesús GARCIA CATALÁN, Carmen MANSO, Joan PETANÁS, Neuus BAENA - (1)Parc Tauli Hospital, Spain
- P16 Paediatric myelin oligodendrocyte glycoprotein antibody-associated disorders: clinical and radiological features
Farah GHARSALLAH, Hanene BENRHOUMA, Abir ZIOUDI, Zouhour MILADI, Thouraya BENYOUNES, Hedia KLAA, Ichraf KRAOUA, Ilhem BENYOUSSEF - (1)Department Of Pediatric Neurology, Lr18sp04, National Institute Mongi Ben Hmida Of Neurology, Tunisia
- P17 Cannabidiol in patients with Dravet Syndrome, Lennox-Gastaut syndrome, and Tuberous Sclerosis: Real-world single-center experience
Marco PERULLI, Maddalena BIANCHETTI, Anna FALCO, Maria PICILLI, Maria Luigia GAMBARDELLA, Michela QUINTILIANI, Ilaria VENEZIA, Ilaria CONTALDO, Chiara VEREDICE, Domenica Immacolata BATTAGLIA - (1)Neuropsychiatria Infantile, Fondazione Policlinico Universitario A. Gemelli, Italy
- P18 A novel WDR45 variant in an encephalopathy mimicking Leigh syndrome with complex I deficiency
Giulia FERRERA (1), Arcangela IUSO (2), Eleonora LAMANTEA (3), Andrea LEGATI (3), Celeste PANTEGHINI (3), Giovanna ZORZI (3), Daniele GHEZZI (3), Anna ARDISSONE (3) - (1)Child Neurology Unit-Department Of Pediatric Neuroscience, Fondazione Ircs Istituto Neurologico Carlo Besta, Italy, (2)Institute Of Neurogenomics, Helmholtz Zentrum München, Italy, (3)Unit Of Medical Genetics And Neurogenetics, Fondazione Ircs Istituto Neurologico Carlo Besta, Italy
- P19 Atypical neuronal ceroid lipofuscinosis type 2 disease: a case report expanding the clinical phenotype
Morgana FREGONESE (1), Federica GRAZIOLA (2), Pierangelo VEGGIOTTI (3), Giovanna ZORZI (2), Federica ZIBORDI (2) - (1)Department Of Biomedical And Clinical Sciences, Postgraduate School Of Child Neuropsychiatry, University Of Milan-Milan (Italy), Italy, (2)Department Of Paediatric Neuroscience, Fondazione Ircs Istituto Neurologico Carlo Besta, 20133, Italy, (3)Biomedical And Clinical Sciences Department, L Sacco, University Of Milan, Italy



POSTERS

- P20 Use of traditionnal, complementary and integrative medicine in the management of pediatric epilepsy at Geneva University Hospitals
Christian KORFF (1) , Celine SAFI (1), Dagmar M. HALLER (2), Mohamed Amir MOUSSA (2) - (1)Geneva University Hospitals, Switzerland, (2)Unige, Switzerland
- P21 Nucleotide Excision Repair (NER) Disorders as differential diagnosis in isolated hypomyelination
Ylenia VAIA (1) , Francesco GAVAZZI (2), Adeline VANDERVER (2), Davide TONDUTI (3) - (1) Unit Of Pediatric Neurology, C.o.a.l.a (center For Diagnosis And Treatment Of Leukodystrophies), V. Buzzi Children's Hospital, Milan, Italy-Università Degli Studi Di Milano - Milan (Italy), Italy, (2) Division Of Neurology, Children's Hospital Of Philadelphia, Philadelphia, Pa, Us, États-Unis, (3) Unit Of Pediatric Neurology, C.o.a.l.a (center For Diagnosis And Treatment Of Leukodystrophies), V. Buzzi Children's Hospital, Milan, Italy-Università Degli Studi Di Milano, Italy
- P22 Mitotic defects in human ASPM microcephaly
Sandrine PASSEMARD (1) , Vincent EL GHOUZZI (2), Jonathan LEVY (3), Sophie LEBON (2), Alexandra ALBERT (2), Renata BASTO (4) - (1)Université Paris Cité, Umr 1141 Neurodiderot, Inserm, Aphp, Hôpital Universitaire Robert Debré, Service De Neurologie Pédiatrique, Paris, France, France, (2)Université Paris Cité, Umr 1141 Neurodiderot, Inserm, Hôpital Universitaire Robert Debré, Paris, France, France, (3)Université Paris Cité, Umr 1141 Neurodiderot, Inserm, Département De Génétique, Hôpital Universitaire Robert Debré, Paris, France, France, (4)Psl Université, Paris, Umr 144 Cnrs, Institut Curie, Paris, France, France
- P23 Rasmussen's encephalitis: A Tunisian pediatric case series
 Meriem BEN HAFSA, Zouhour MILADI, Thouraya BEN YOUNES, Maha JAMOUSSEI, Abir ZIOUDI, Hedia KLAA, Ichraf KRAOUA, Hanene BENRHOUMA, Ilhem BEN YOUSSEF-TURKI - (1)Research Unit Lr18sp04 And Department Of Child And Adolescent Neurology, National Institute Mongi Ben Hmida Of Neurology Of Tunis, Tunisia
- P24 POLIII-related leukodystrophy: clinical, imaging and genetic findings in six Tunisian families
Ichraf KRAOUA (1) , Maha JAMOUSSEI (1), Cyrine DRISSI (2), Lilia KRAOUA (3), Hanene BENRHOUMA (1), Thouraya BEN YOUNES (1), Sonia NAGI (2), Sonia ABDELHAK (4), Odile BOESPFLUG TANGUY (5), Ilhem BEN YOUSSEF TURKI (1), Mediha TRABELSI (3), Imen DORBOZ (5) - (1)Lr18sp04, Department Of Child And Adolescent Neurology, National Institute Mongi Ben Hmida Of Neurology, Faculty Of Medicine Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (2)Department Of Neuroradiology, National Institute Mongi Ben Hmida Of Neurology, Faculty Of Medicine Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (3) Department Of Congenital And Hereditary Diseases, Charles Nicolle Hospital, Faculty Of Medicine Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (4)Lr11pt05, Laboratory Of Biomedical Genomics And Oncogenetics, Pasteur Institute Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (5)Université De Paris, Neurodiderot, Umr 1141, Inserm, Neuropédiatrie, Leukofrance, Aphp, Hôpital Robert Debré, France., France



POSTERS

- P25 **Megalencephalic leukoencephalopathy with subcortical cysts: a report of five Tunisian families**
Maha JAMOUSSI (1), Hanene BENRHOUMA (1), Thouraya BEN YOUNES (1), Meriem BEN HAFSA (1), Sonia ABDELHAK (2), Odile BOESPFLUG TANGUY (3), Ilhem BEN YOUSSEF TURKI (1), Imen DORBOZ (3), Ichraf KRAOUA (1) - (1)Lr18sp04, Department Of Child And Adolescent Neurology, National Institute Mongi Ben Hmda Of Neurology, Faculty Of Medicine Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (2)Lr11pt05, Laboratory Of Biomedical Genomics And Oncogenetics, Pasteur Institute Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (3)Université De Paris, Neurodiderot, Umr 1141, Inserm, Neuropédiatrie, LeukoFrance, Aphp, Hôpital Robert Debré, France, France
- P26 **Aicardi Goutieres syndrome: study of a Tunisian cohort**
Maha JAMOUSSI (1), Thouraya BEN YOUNES (1), Hanene BENRHOUMA (1), Meriem BEN HAFSA (1), Sonia ABDELHAK (2), Odile BOESPFLUG TANGUY (3), Ilhem BEN YOUSSEF TURKI (1), Imen DORBOZ (3), Ichraf KRAOUA (1) - (1)Lr18sp04, Department Of Child And Adolescent Neurology, National Institute Mongi Ben Hmda Of Neurology, Faculty Of Medicine Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (2)Lr11pt05, Laboratory Of Biomedical Genomics And Oncogenetics, Pasteur Institute Of Tunis, University Of Tunis El Manar, Tunis, Tunisia, (3)Université De Paris, Neurodiderot, Umr 1141, Inserm, Neuropédiatrie, LeukoFrance, Aphp, Hôpital Robert Debré, France, France
- P27 **Developmental and epileptic encephalopathy linked to UFSP2 gene mutations**
Ana ULINICI, Emanuelle RANZA, Jasmine HIRSCH, Mary KURIAN - (1)Hopital Cantonal Fribourg, Switzerland
- P28 **Psychomotor regression in migrant children**
Florian PERRIN DE BRICHAMBAUT (1), Vincent PARGNY (2), Rebecca MORÉ (1), Hortense PETAT (2), Isabelle MICHELET (2), Yacine ZEGGAY (3), Marie Cardine AUDE (4), Stéphane MARRET (1), Ivana DABAJ (1) - (1)Department Of Neonatology, Pediatric Intensive Care, And Pediatric Neurology, Rouen University Hospital, France, (2)Pediatric Department, Rouen University Hospital, France, (3)Department Of Infection And Tropical Diseases, Rouen University Hospital, France, (4)Pediatric Hematology-Oncology Department, Rouen University Hospital, France
- P29 **Spongy degeneration of CNS in Canavan Disease: links between onset and severity**
Alina ANDREI (1), Lorraina CHAVÉE (1), Grammatina BOITSIOS (2), Luc REGAL (3), Anneleen MORTIER (4), Bavo BLOMME (4), Catheline VILAIN (5), Robert MLLER (6), Alec AEBY (1) - (1)Department Of Pediatric Neurology, Hôpital Universitaire De Bruxelles (h.u.b), Belgium, (2) Radiology Unit, Hôpital Universitaire De Bruxelles (h.u.b), Belgique, (3)Department Of Metabolic Diseases, Hôpital Universitaire De Bruxelles (h.u.b), Belgique, (4)Department Of Clinical Chemistry And Radioimmunology, Laboratory For Metabolic Diseases, Uz Brussels, Belgique, (5)Department Of Genetics, Hôpital Universitaire De Bruxelles (h.u.b), Belgium, (6)Department Of Paediatrics, Ch Mouscron, Belgium



POSTERS

- P30 Altered DNA methylation and gene expression predict disease severity in patients with Aicardi-Goutières syndrome
Davide POLITANO (1), Jessica GARAU (2), Amandine CHARRAS (3), Costanza VARESIO (1), Francesca DRAGONI (4), Jessica GALLI (5), Stella GAGLIARDI (6), Orietta PANSARASA (7), Elisa FAZZI (5), Simona ORCESI (1), Cristina CEREDA (8), Christian M. HEDRICH (9) – (1)Department Of Brain And Behavioral Sciences, University Of Pavia-Pavia (Italy); Child Neurology And Psychiatry Unit, Irccs Mondino Foundation, Italy, (2)Neurogenetics Research Centre, Irccs Mondino Foundation, Italy, (3)Department Of Women's And Children's Health, Institute Of Life Course And Medical Sciences, University Of Liverpool, United Kingdom, (4)Department Of Biology And Biotechnology, University Of Pavia – Pavia (Italy); Molecular Biology And Transcriptomics, Irccs Mondino Foundation, Italy, (5)Department Of Clinical And Experimental Sciences, University Of Brescia – Brescia (Italy); Unit Of Child Neurology And Psychiatry, Asst Spedali Civili Of Brescia, Italy, (6)Molecular Biology And Transcriptomics, Irccs Mondino Foundation, Italy, (7)Cellular Model And Neuroepigenetics, Irccs Mondino Foundation, Italy, (8)Functional Genomic And Rare Diseases Unit, Department Of Pediatrics, Children's Hospital "v. Buzzi", Italy, (9)Department Of Women's And Children's Health, Institute Of Life Course And Medical Sciences, University Of Liverpool-Liverpool, (United Kingdom); Department Of Paediatric Rheumatology, Alder Hey Children's Nhs Foundation Trust Hospital, United Kingdom
- P31 When cognitive disorder reveals motor disorder and conversely ?
Sibylle GONZALEZ MONGE, Véronique MANEL – (1)Hospices Civils De Lyon, France
- P32 Headache with Neurologic Deficits and cerebrospinal fluid Lymphocytosis (HaNDL) syndrome in latent HHV6 infection
Anna FETTA (1), Diana LA MALFA (2), Carlotta BIAGI (3), Caterina CAMPOLI (4), Marcello LANARI (3), Duccio Maria CORDELLI (1) – (1)Irccs Istituto Delle Scienze Neurologiche Di Bologna, Uoc Neuropsichiatria Dell'età Pediatrica-Department Of Medical And Surgical Sciences, University Of Bologna, Italy, (2)Specialty School Of Paediatrics, University Of Bologna, Italy, (3)Pediatric Emergency Unit, Irccs Azienda Ospedaliero-Universitaria Di Bologna, Italy, (4) Infectious Diseases Unit, Department For Integrated Infectious Risk Management, Irccs Azienda Ospedaliero-Universitaria Di Bologna, Italy
- P33 Phenotypic spectrum of COL4A1/A2 related disorders: what can we learn from literature?
Eleonora BONAVENTURA (1), Daria MARELLI (2), Simona ORCESI (3), Davide TONDUTI (1) – (1)Child Neurology Unit, V. Buzzi Children's Hospital, Italy, (2)University Of Milan, Italy, (3)Child Neurology And Psychiatry Unit, Irccs Mondino Foundation, Italy
- P34 Abnormal eye movements in an infant with SARS-COV2 infection, a possible case of parainfective opsoclonus?
Silvia BENEDETTI, Eldahaby DANIELA, Elisa SCHIAVON, Emilia RICCI, Maria Paola CANEVINI – (1)Unimi, Italy



POSTERS

- P35 Perinatal stroke: risk factors, clinical presentation and neurodevelopmental outcome in term and late-preterm neonates
Federica TEUTONICO (1), Anna FREDDI (2), Paola DONEDA (1), Roberta RESTELLI (1), Stefano MARTINELLI (1), Aglaia VIGNOLI (3) - (1)Niguarda Hospital, Italy, (2)University Of Milan, Italy, (3)Niguarda Hospital, University Of Milan, Italy
- P36 Rehabilitation perspectives in Landau-Kleffner Syndrome: Sign Language as an effective tool in addressing severe verbal auditory agnosia
Giusi FERRARA (1), Luca ANDREOLI (2), Maria Gaia REDAELLI (3), Valeria LEONARDI (4), Elisa GRANOCCHIO (3), Silvia ESPOSITO (3), Santina MAGAZU' (3), Pierangelo VEGGIOTTI (5), Stefania Maria BOVA (6) - (1) Child And Adolescent Neuropsychiatry Unit, Asst Fatebenefratelli Sacco, Italy, (2) Department of Biomedical and Clinical Sciences, Postgraduate School of Child Neuropsychiatry, University of Milan, Milan, Italy, (3) Developmental Neurology Unit, Fondazione Irccs Istituto Neurologico Carlo Besta, Italy, (4) Department Of Biomedical And Clinical Sciences, L. Sacco, University Of Milan, Italy, (5) Child Neurology Unit, Buzzi Children's Hospital, University Of Milan, Italy, (6) Child Neurology Unit, Buzzi Children's Hospital, Italy
- P37 PRRT2-associated disease: the spectrum of clinical features observed in our centre
Lara ADAMI, Elisabetta FIORELLI, Elisabetta MARROCCO, Ylenia VAIA, Roberto PREVITALI, Eleonora BONAVENTURA, Monica LODI, Enrico ALFEI, Davide TONDUTI, Pierangelo VEGGIOTTI - (1)Pediatric Neurology Unit, V. Buzzi Children's Hospital, Asst Fatebenefratelli Sacco - Università Degli Studi Di Milano, Milan, Italy
- P38 Relapse neurological deficits (facilitated by viral infections): when a pseudo-inflammatory presentation reveals a genetic disease
Florence RENALDO (1), Mohamed Khayri MSALBI (2), Boris KEREN (3), Gilles BRETAUDEAU (4), Hélène RAUSCENT (4), Claudia RAVELLI (1), Diane DOUMMAR (1), Cyril MIGNOT (5), Diana RODRIGUEZ (6) - (1)Department Of Paediatric Neurology, Center For Rare Neurogenetic Diseases, Armand Trousseau-La Roche Guyon Childrens' Hospital, Ap-Hp, Sorbonne University, Paris, France, (2)Service De Pédiatre Ch De Gonesse 95500, France, (3)Ap-Hp, Hôpital De La Pitié-Salpêtrière, Département De Génétique, F-75013 Paris, France, France, (4)Service De Pédiatrie Chu Hôpital Rennes Cedex 2, France, (5)Sorbonne Université, Département De Génétique, Groupe Hospitalyr Pitié-Salpêtrière And Hôpital Trousseau, Centre De Référence Déficiences Intellectuelles De Causes Rares, Paris, France, (6)Department Of Paediatric Neurology, Center For Rare Neurogenetic Diseases, Armand Trousseau-La Roche Guyon Childrens' Hospital, Ap-Hp, Sorbonne University, Paris, France
- P39 Panayiotopoulos syndrome vs viral encephalitis: a case report
Francesca SILINI (1), Eleonora MORABITO (1), Emanuele LABRUNA (1), Stefania Maria BOVA (2), Monica LODI (2), Pierangelo VEGGIOTTI (3) - (1)Department Of Biomedical And Clinical Science, University Of Milan, Milan, Italy, (2)Pediatric Neurology Unit, V. Buzzi Children's Hospital, Asst Fatebenefratelli Sacco, Milan, Italy, (3)Department Of Biomedical And Clinical Science, University Of Milan, Milan Italy, Pediatric Neurology Unit, V. Buzzi Children's Hospital, Asst Fatebenefratelli Sacco, Milan, Italy



POSTERS

- P40 **Expanding the clinical phenotype of NMNAT1-related disorders**
Roberta NICOTRA (1), Federica MORELLI (2), Antonella LUPARIA (3), Daria PAINI (3), Chiara BERTONE (4), Giulio RUBERTO (3), Anna PICCHIECCHIO (5), Davide TONDUTI (6), Sabrina SIGNORINI (7) – (1) Department Of Child Neurology And Psychiatry, Irccs Mondino Foundation, Pavia, Italy (2) Department Of Brain And Behavioral Sciences, University Of Pavia, Pavia, Italy, Italy, (2) Department Of Brain And Behavioral Sciences, University Of Pavia, Pavia, Italy (3) Developmental Neuro-Optthalmology Unit, Irccs Mondino Foundation, Pavia, Italy, (3) Developmental Neuro-Optthalmology Unit, Irccs Mondino Foundation, Pavia, Italy, (4) Department Of Surgical And Clinical, Diagnostic And Pediatric Sciences, Section Of Ophthalmology, Irccs Fondazione Policlinico San Matteo, University Of Pavia, 27100 Pavia, Italy, (5) Department Of Brain And Behavioral Sciences, University Of Pavia, Pavia, Italy, (6) Neuroradiology Department, Irccs Mondino Foundation, 27100 Pavia, Italy, Italy, (6) Unit Of Pediatric Neurology, C.o.a.l.a. (center For Diagnosis And Treatment Of Leukodystrophies), V. Buzzi Children's Hospital, Università Degli Studi Di Milano, Milan, Italy, (7) Department Of Child Neurology And Psychiatry, Irccs Mondino Foundation, Pavia, Italy; 3 Developmental Neuro-Optthalmology Unit, Irccs Mondino Foundation, Pavia, Italy
- P41 **Clinical Characteristics and Diagnostic Features of Guillain-Barré Syndrome Among the Pediatric Population**
Germana LO MONACO (1), Elisabetta FIORELLI (1), Arianna GADDA (1), Matilde FERRARIO (2), Laura FIORI (2), Paola ERBA (2), Francesca REDAELLI (2), Valentina FABIANO (3), Anna MANDELLI (4), Matteo PESSINA (4), Chiara DONEDA (5), Emilio CIUSANI (6), Diego FRANCIOTTA (7), Matteo GASTALDI (7), Maurizio OSIO (8), Gian Vincenzo ZUCCOTTI (9), Pierangelo VEGGIOTTI (10), Eleonora BONAVENTURA (11), Stefania Maria BOVA (11) – (1)Department Of Biomedical And Clinical Science, University Of Milan, Italy, (2)Department Of Pediatric, “vittore Buzzi” Children's Hospital, Asst-Fbf-Sacco, Italy, (3)Department Of Pediatric, “vittore Buzzi” Children's Hospital, Asst-Fbf-Sacco, Department Of Biomedical And Clinical Science, Italy, (4)Pediatric Intensive Care, “vittore Buzzi” Children's Hospital, Italy, (5) Department Of Pediatric Radiology And Neuroradiology, V. Buzzi Children's Hospital, Italy, (6) Department Of Research And Technology, Fondazione Irccs Istituto Neurologico C. Besta, Italy, (7)Neuroimmunology Research Unit, Irccs Mondino Foundation, Pavia, Italy, Italy, (8)Department Of Neurology Asst Fatebenefratelli Sacco, Italy, (9)Department Of Pediatric, “vittore Buzzi” Children's Hospital, Asst-Fbf-Sacco, Department Of Biomedical And Clinical Science, University Of Milan, Italy, (10)Pediatric Neurology Unit, Vittore Buzzi Children's Hospital, department Of Biomedical And Clinical Science, University Of Milan, Italy, (11)Pediatric Neurology Unit, Vittore Buzzi Children's Hospital, Italy



POSTERS

- P42** Beyond CADASIL Syndrome: biallelic NOTCH3 variants in a paediatric patient with leukoencephalopathy, brain calcifications, acquired microcephaly, hemiparesis and autism spectrum disorder
Marina MARTINEZ POPPLE (1), Laura SIRI (1), Francesca FARAVELLI (2), Maria Francesca DI FEO (2), Francesca MADIA (3), Mariasavina SEVERINO (4), Deborah PREITI (5), Andrea ROSSI (6), Lino NOBILI (7), Elisa DE GRANDIS (7) – (1)Child Neuropsychiatry Unit, Irccs Istituto Giannina Gaslini, Italy, (2)Genomics And Clinical Genetics Unit, Irccs Istituto Giannina Gaslini, Italy, (3)Medical Genetics Unit, Irccs Istituto Giannina Gaslini, Italy, (4)Neuroradiology Unit, Irccs Istituto Giannina Gaslini, Italy, (5)Psychology Unit, Irccs Istituto Giannina Gaslini, Italy, (6)Neuroradiology Unit, Irccs Istituto Giannina Gaslini; Department Of Health Sciences (dissal), University Of Genova, Italy, (7)Child Neuropsychiatry Unit, Irccs Istituto Giannina Gaslini; Department Of Neurosciences, Rehabilitation, Ophthalmology, Genetics And Mother–Child Health (dinogmi), University Of Genova, Italy
- P43** Recurrent Arterial Ischemic stroke in a 8-year-old girl from Mali
Amadou Mahamane TOURE (1), Alassane Baneye MAIGA (2), Adama KONÉ (3), Mariame DIAMOUTÉNÉ (3), Andoule GUINDO (3), Mahamadou TRAORÉ (3), Karamoko SACKO (1), Hawa KONARÉ (1), Abdoul Karim DOUMBIA (1), Belco MAIGA (1), Guida LANDOURE (2) – (1)Chu Gabriel Toure, Mali, (2)Chu Point G, Mali, (3)Clinique Kaidara, Mali
- P44** Cerebellar grey matter volumes correlate with cognitive scores in patients with paediatric Opsoclonus–Myoclonus–Ataxia Syndrome
Marina MARTINEZ POPPLE (1), Mariasavina SEVERINO (2), Deborah PREITI (3), Massimo CONTE (4), Angela PISTORIO (5), Domenico TORTORA (2), Laura SIRI (1), Andrea ROSSI (6), Lino NOBILI (7), Elisa DE GRANDIS (7) – (1)Child Neuropsychiatry Unit, Irccs Istituto Giannina Gaslini, Italy, (2)Neuroradiology Unit, Irccs Istituto Giannina Gaslini, Italy, (3)Psychology Unit, Irccs Istituto Giannina Gaslini, Italy, (4)Paediatric Haematology And Oncology Unit, Irccs Istituto Giannina Gaslini, Italy, (5)Scientific Department, Biostatistics Unit, Irccs Istituto Giannina Gaslini, Italy, (6)Neuroradiology Unit, Irccs Istituto Giannina Gaslini; Department Of Health Sciences (dissal), University Of Genova, Italy, (7)Child Neuropsychiatry Unit, Irccs Istituto Giannina Gaslini; Department Of Neurosciences, Rehabilitation, Ophthalmology, Genetics And Mother–Child Health (dinogmi), University Of Genova, Italy
- P45** Retinal degeneration and intellectual disability in a patient with pathogenic variants in SCL6A6 gene: a case report
Roberta NICOTRA (1), Federica MORELLI (2), Gloria ROSSETTO (1), Enza Maria VALENTE (3), Simone GANA (4), Mauro ANTONINI (4), Elena SALIGARI (5), Sabrina SIGNORINI (6), Walter MISEFARI (6) – (1)1 Department Of Child Neurology And Psychiatry, Irccs Mondino Foundation, Pavia, Italy, (2)2 Department Of Brain And Behavioral Sciences, University Of Pavia, Pavia, Italy, (3)3 Developmental Neuro–Ophthalmology Unit, Irccs Mondino Foundation, Pavia, Italy, (4)4 Department Of Molecular Medicine, University Of Pavia, Pavia, Italy, (5)5 Neurogenetics Research Unit, Irccs Mondino Foundation, Pavia, Italy, (6)6 Department Of Molecular Medicine, University Of Pavia, Pavia, Italy, (7)3 Developmental Neuro–Ophthalmology Unit, Irccs Mondino Foundation, Pavia, Italy, (6)1 Department Of Child Neurology And Psychiatry, Irccs Mondino Foundation, Pavia, Italy; 3 Developmental Neuro–Ophthalmology Unit, Irccs Mondino Foundation, Pavia, Italy



POSTERS

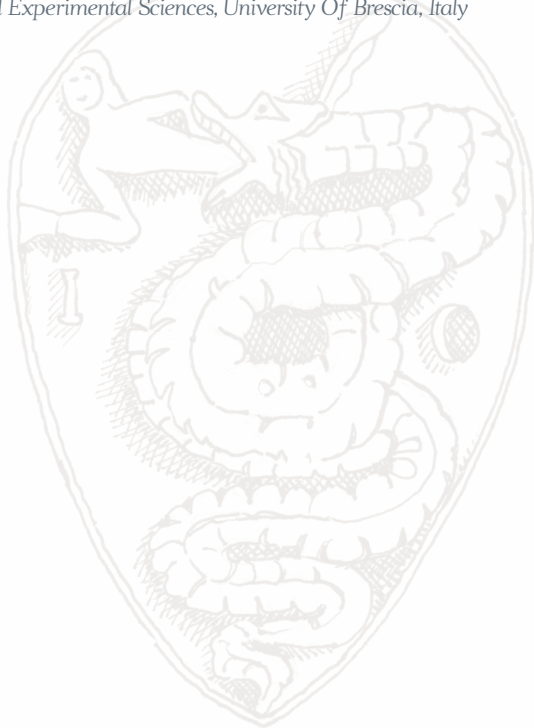
- P46 **Neurovisual profile in Dravet syndrome: prospective longitudinal study from infancy to adulthood**
Chiara PORTO, Giuseppe MARSELLA, Giovanna MASONE IACOBUCCHI, Maria PETRIANNI, Lorenzo ORAZI, Michela QUINTILIANI, Maria Luigia GAMBARDELLA, Daniela Pia Rosaria CHIEFFO, Daniela RICCI, Domenica Immacolata BATTAGLIA - (1)Irccs Fondazione Policlinico Universitario Agostino Gemelli, Italy
- P47 **A case of acute encephalopathy in a 12-year-old girl with de novo mutation in the DNMI1 gene: suggestions for early detection and differential diagnosis**
Chiara ZACCHE (1), Maria Luigia GAMBARDELLA (2), Michela QUINTILIANI (2), Marco PERULLI (2), Silvia PULITANO (2), Ida TURRINI (2), Domenica Immacolata BATTAGLIA (2), Ilaria CONTALDO (2), Chiara VEREDICE (2) - (1)Pediatric Neurology Unit, Università Cattolica Del Sacro Cuore, Italy, (2)Pediatric Neuropsychiatric Unit, Dipartimento Di Salute Della Donna E Del Bambino E Sanità Pubblica, Fondazione Policlinico Universitario Agostino Gemelli, Irccs, Italy
- P48 **Delineating the phenotypic spectrum of the recurrent monoallelic mutation of the TMEM106B gene: report of 3 new patients**
Marie-Aricie VEKEMANS (1), François CHALARD (1), Pierre BEZE-BEYRIEC (2), Sebastien CABASSON (2), Mathieu MILH (3), Matthieu ROBERT (4), Desnoux BEATRICE (3), Girard NADINE (5), Renaldo FLORENCE (1), Burglen LYDIE (1), Rodriguez DIANA (1) - (1)Centre De Référence Neurogénétique & Service De Neuropédiatrie, Hôpital Armand-Trousseau, Aphp, Sorbonne Université, France, (2)Service De Pédiatrie, Centre Hospitalier De Pau, France, (3)Service De Neuropédiatrie, Hôpital De La Timone-Enfants, Aphp, Aix-Marseille Univ, France, (4)Service D'ophtalmologie Pédiatrique, Hôpital Necker Enfants Malades, Aphp, Université Paris Cité, France, (5)Service De Radiologie, Hôpital De La Timone-Enfants, Aphp, Aix-Marseille Univ, France
- P49 **Comparison between an atypical and a typical case of Subacute Sclerosing Panencephalitis: when to suspect this rare but still present condition**
Chiara BAGLIANI (1), Luca BOSISIO (1), Marta NEBIOLO (2), Maria Margherita MANCARDI (1), Edoardo CANALE (1), Domenico TORTORA (3), Laura SIRI (1), Pasquale STRIANO (2), Lino NOBILI (1), Elisabetta AMADORI (1) - (1)Unit Of Child Neuropsychiatry, Irccs Giannina Gaslini, Ern Epicare Network, Genoa, Italy 2department Of Neurosciences, Rehabilitation, Ophthalmology, Genetics And Maternal And Child Health (dinogmi), University Of Genoa, Italy, (2)Unit Of Pediatric Neurology And Muscular Diseases, Irccs Giannina Gaslini, Ern Epicare Network, Genoa, Italy, (3)Unit Of Neuroradiology, Irccs Giannina Gaslini, Genoa, Italy
- P50 **The continuum between Down Syndrome Disintegrative Disorder and Autoimmune Encephalitis: diagnostic and therapeutic considerations**
Beatrice BELLINI (1), Anna MOLINARO (2), Jessica GALLI (2), Sara BRUNETTI (3), Lucio GIORDANO (3), Elisa FAZZI (2) - (1)Clinical And Experimental Sciences Department, University Of Brescia, Italy, (2)Clinical And Experimental Sciences Department, University Of Brescia; Children And Adolescent Neurology And Psychiatry Unit, Asst Spedali Civili, Italy, (3)Children And Adolescent Neurology And Psychiatry Unit, Asst Spedali Civili, Italy



POSTERS

P51 The nature and frequency of Developmental Coordination Disorders in children with Neurofibromatosis Type 1

Serena MICHELETTI (1) , Anna MOLINARO (2), Giorgio BONOMI (3), Flavio Cesar QUESQUEN SALDOVAL (3), Elisa FAZZI (2) - (1)Unit Of Child Neurology And Psychiatry, Asst Spedali Civili Of Brescia, Italy, (2)Unit Of Child Neurology And Psychiatry, Asst Spedali Civili Of Brescia, Department Of Clinical And Experimental Sciences, University Of Brescia, Italy, (3)Department Of Clinical And Experimental Sciences, University Of Brescia, Italy



SYMPOSIUMS / SYMPOSIA

Vendredi 17 mars / Friday, March 17



Rare and Complex Epilepsies: Focus on Comorbidities and Cognitive Issues

Moderator: Pierangelo Veggiotti

- Comorbidities in Rare and Complex Epilepsies
Pasquale Striano
- Exploring Complexities: Cognitive Issues in Children with Epilepsy
Stefania Maria Bova



Seizure and Beyond: The Management of DEEs

Moderator: Aglaia Vignoli

- Cannabidiol: a new treatment opportunity in DEEs
Francesca Ragona
- Clinical Cases presentation
Roberto Previtali, Silvia Masnada



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15 mars | March 15

19h30 | 7.30 pm

Via San Vittore 21, 20123 Milan

Diner de Gala

(non inclus dans les frais d'inscription)

Le dîner de gala se tiendra au Musée des sciences et des techniques Léonard de Vinci, dans la salle « Cenacolo ». (Dîner payant sous réserve d'inscription au préalable).

Gala Dinner

(not included in the registration fees)

The Gala Dinner will be held in the Leonardo da Vinci Museum of Science and Technology, in the "Cenacolo" Hall.

(Paying dinner upon registration)

15 mars | March 15

20h30 | 8.30 pm

Via San Vittore 21, 20123 Milan



A circular diagram with a crosshair and a compass rose, likely representing a celestial or navigational instrument. The diagram features a central cross with four arms, and a circular frame containing various lines and markings, possibly representing a celestial sphere or a navigational chart. The style is that of a technical drawing or a historical illustration.

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