CONSENSUS CONFERENCE

AICARDI SYNDROME:

FROM DEFINING THE PHENOTYPE TO UNRAVELLING THE GENOTYPE

Aicardi syndrome is a rare disease defined classically by the occurrence of congenital chorioretinal lacunae, corpus callosum dysgenesis and epileptic spasms. The syndrome shows a dominant transmission apparently linked to the X chromosome, but its cause is unknown. Because of the absence of a specific genetic hallmark, diagnosis is based on clinical features. While the simultaneous presence of the three classical features permits an unquestionable diagnosis, a relatively high number of patients present with only two of the classical features. Therefore, modified diagnostic criteria have also been proposed, which require the presence of two classical features plus other major or supporting features . Lack of general consensus on diagnostic criteria, coupled with variability in clinical and EEG phenotypes, is one of the factors that complicate research into the genetic mechanisms responsible for the manifestations of the syndrome. This background provides the rationale for the present workshop, which will bring together an international group of experts with a special interest in the clinical, neurophysiological and neuroradiological manifestations of Aicardi syndrome, and in the genetic factors involved in its pathogenesis. The overall purpose is to share personal experiences with respect to the diagnosis and management of the syndrome, to achieve consensus on a minimal set of diagnostic criteria, to exchange findings from ongoing research, and to set the basis for a collaborative effort aimed at unravelling the underlying etiology.

Friday November 16th 2018
(Closed Workshop)

9:00-9:05 Welcome and introduction
Emilio Perucca (Pavia)

SESSION 1. OVERVIEW ON CLINICAL FEATURES AND DIAGNOSTIC CRITERIA OF AICARDI SYNDROME
Chairs: Nadia Bahi-Buisson (Paris) and Pierangelo Veggiotti (Milan)

9:05-10:05 New results on clinical, electroclinical and neuroradiological aspects
Mara Cavallin (Paris) and Silvia Masnada (Pavia)

10:05-12:30 General discussion: Towards a consensus in redefining diagnostic criteria
Facilitators: Nadia Bahi-Buisson (Paris) and Pierangelo Veggiotti (Milan)

12:30-13:30 Management issues (including discussion)
Alexis Arzimanoglou (Lyon)

13:30-14:30 Lunch

SESSION 2. GENETIC STUDIES AND NEW PERSPECTIVES
Chairs: Jozef Gecz (Adelaide) and Igna van der Veyver (Houston)

14:30-16:30 Presentation of ongoing genetic studies from the Australian, French and Italian collaborative Groups
Mark Corbett (Adelaide), Federico Zara (Genoa), Igna van der Veyver (Houston), Mara Cavallin (Paris), Elliot Sherr (San Francisco)

16:30- 16:45 Coffee Break

16:45-19:30 General discussion
Unravelling the genetics of Aicardi syndrome: Optimization of research strategies and discussion of potential collaborations

FONDAZIONE MONDINO
Istituto Neurologico Nazionale a Carattere Scientifico | IRCCS

European Reference Network for rare or low prevalence complex diseases — Network Epilepsies (ERN EpiCARE)

EpiCARE

HOW ADVANCES IN RESEARCH ARE CHANGING THE DIAGNOSTIC AND THERAPEUTIC LANDSCAPE IN EPILEPSY

November 17th, 2018 - IRCCS Mondino Foundation (Berlucchi & Rossi Hall), Pavia

9:00-9:30 Registration of participants

9:30-9:40 Welcome and introduction
Fabio Blandini and Emilio Perucca (Pavia)

Session 1. Breakthroughs in genetic research and clinical applications*

Chairs: Alexis Arzimanoglou (Lyon) and Enza Maria Valente (Pavia)

9:40-10:10 Chromosome X and epilepsy
Jozef Getz (Adelaide)

10:10-10:40 Epilepsy genetics: beyond Mendel and coding mutations
Mark Corbett (Adelaide)

10:40-11:10 Brain development in the epilepsies
Nadia Bahi-Buisson (Paris)

11:10-11:30 Coffee break

11:30-12:00 Epileptic encephalopathies as genetic disorders – transmitted or acquired?
Elliott Sherr (San Francisco)

12:00-12:30 Doctor, what is the cause of my epilepsy? When and how to apply genetic testing in the routine clinical setting
Federico Zara (Genoa)

12:30-13:00 Prenatally detected CNS defects and genetic syndromes
Igna Van den Veyver *(Houston)*

13:00-14:00 Lunch break

Session 2. Recent and future therapeutic breakthroughs**

Chairs: Carlo Andrea Galimberti (Pavia), Pierangelo Veggiotti (Milan)

14:00-14:30 From genetic testing to precision therapies for epilepsies
Renzo Guerrini (Florence)

14:30-15:00 Cannabis in epilepsy: What have we learnt from controlled trials, and what is next?
Emilio Perucca (Pavia)

15:00-15:30 Translating genetic epilepsy research into innovative non-pharmacological therapeutic approaches
Vania Broccoli (Milan)

15:30-16:00 How biomarkers are changing our approach to discovery and testing of antiepileptogenic drugs
Annamaria Vezzani (Milan)

16:00-16:30 The Aicardi syndrome collaboration network: Clinical, neuro-radiological and therapeutic aspects
Silvia Masnada (Pavia) Mara Cavallin (Paris)

16:30-17:00 CME test and closing

* *Session in English / ** Session in Italian*

17:00-18:00 Closed meeting with Aicardi Association families

Aims of the Symposium

During the last 20 years there have been impressive advances in our understanding of the mechanisms underlying common and rare epileptic syndromes, particularly in the area of genetics. The objective of the present Conference is to highlight some key areas where findings from genetic research have led to a truly major improvement in knowledge about mechanisms involved in brain development and in the pathogenesis of several forms of epilepsy with onset in infancy and childhood. Special emphasis will be placed on practical aspects, including the need to refine diagnostic criteria, to improve the efficiency of genetic testing, and to address implications for clinical management. Unravelling the molecular mechanisms of epilepsies is already leading to the development and application of new and more effective precision therapies which target directly the etiology of the disease rather than merely its symptoms. The Conference will also address other recent advances in therapeutic research, from recently introduced plant-derived compounds to biomarker-driven disease-modifying interventions and to innovative non-pharmacological treatments.

Scientific Committee

Valentina De Giorgis, Silvia Masnada, Emilio Perucca, Pierangelo Veggiotti and Federico Zara

Coordination and registration

Silvia Molinari
Formazione&Informazione Office, IRCCS Fondazione Mondino , Pavia - Email: formazione.informazione@mondino.it

On line registration at: http://corsi.mondino.it/corsi_list.php

CME-CPD

The Conference has been accredited by Regione Lombardia for Continuing Medical Education with 6 (six) ECM credits for:

Medico Chirurgo (Biochimica clinica, Genetica Medica, Laboratorio di Genetica Medica, Medicina Generale-Medici di Famiglia, Neurofisiopatologia, Neurologia, Neuropsichiatria infantile, Neuroradiologia, Ortopedia e Traumatologia, Pediatria, Pediatria-Pediatri di libera scelta, Radiodiagnostica), **Fisioterapista, Biologo; Infermiere; Infermiere Pediatrico, Psicologo** (Psicologia, Psicoterapia)**, Tecnico di Neurofisiopatologia, Tecnico Sanitario di Radiologia Medica, Terapista della Neuro e Psicomotricità dell'Età Evolutiva, Terapista Occupazionale.**

To obtain credits, participants must attend the whole conference and pass the evaluation test by answering correctly at least 80% of the questions.

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