

FRIDAY OCTOBER 10 – VENDREDI 10 OCTOBRE

ROOM THEATRE / SALLE THEATRE

SATELLITE SYMPOSIUM / SYMPOSIUM SATELLITE

8.30 - 9.45

REGISTRATION / ACCUEIL INSCRIPTIONS

European and Rare Diseases: Hoping to Meet Patients and Families Expectations

L'Europe et les maladies rares : un espoir à la mesure des attentes des malades et des familles

LANGUAGE: FRENCH (SIMULTANEOUS TRANSLATION INTO ENGLISH)

LANGUE : FRANÇAIS (TRADUCTION SIMULTANÉE EN ANGLAIS)

Under the High Patronage of Mister Nicolas Sarkozy, President of the French Republic

Organized by Alliance Maladies Rares

In partnership with AFM (French Association against Muscular Dystrophy)

In collaboration with EURORDIS & AFSR

Sous le Haut Patronage du Président de la République Française, Monsieur Nicolas Sarkozy

Organisé par l'Alliance Maladies Rares

En partenariat avec l'AFM

En collaboration avec EURORDIS et l'AFSR

9.45

Message from the President of the French Republic, Mister Nicolas Sarkozy

Message du Président de la République Française, Monsieur Nicolas Sarkozy

Introduction / Introduction

Françoise Grossetête, European Parliament / Parlement Européen

10.00 - 11.15

SESSION 1

RARE DISEASES: A PUBLIC HEALTH PRIORITY IN EUROPE?

LES MALADIES RARES : UNE PRIORITÉ DE SANTÉ PUBLIQUE DE L'EUROPE ?

Chair / Modérateur : Francis Brunelle, Necker Hospital, AP-HP / Hôpital Necker, AP-HP (Assistance Publique-Hôpitaux de Paris)

The French Experience, a Source of Inspiration for Europe?

L'expérience française : une source d'inspiration pour l'Europe ?

Ségolène Aymé, Rare Disease Task Force

Rare Diseases in Europe: Recent Advances and Perspectives

Les maladies rares et l'Europe : avancées récentes et perspectives

Toni Montserrat, DG SANCO, European Commission / DG SANCO, Commission Européenne

Orphan Drugs, a Policy Initiative Success

Les médicaments orphelins : une initiative politique réussie

Agnès de Saint Raymond, EMEA, European Medicines Agency / EMEA, Agence Européenne du Médicament

11.15 - 12.45

SESSION 2

RARE DISEASES AND RESEARCH POLICY : EUROPE A MANDATORY LINK

LES MALADIES RARES ET LA RECHERCHE : L'EUROPE, UN ÉCHELON INDISPENSABLE

Chair / Modérateur : Dominique Daegelen, Institut Génétique et Développement, INSERM

Research Policy on Rare Diseases: Lessons from French Experience

La recherche sur les maladies rares : les leçons tirées de l'expérience française

Elisabeth Tournier-Lasserre, GIS Rare Diseases Institute / GIS-Institut Maladies Rares



A Hand Link from Gene to Care

Rare Diseases, a Research Priority for Europe?

Les maladies rares : une priorité de recherche pour l'Europe ?

Catherine Berens, DG Research, European Commission / DG Recherche, Commission Européenne

The European Dimension Interest for Research:

L'intérêt de la dimension européenne pour la recherche :

- Sharing of Expertise and Resources
- **une complémentarité de compétences et de ressources**

Laurent Villard, INSERM

- An Added Value for Clinical Trials
- **Une plus-value pour les essais cliniques**

Nicolas Levy, INSERM

12.45 - 14.30 BREAK / PAUSE

14.30 - 15.45 **SESSION 3**

FOR A BEST MANAGEMENT OF PATIENTS: NEEDS FOR AN EUROPEAN LEVEL ORGANISATION

POUR UNE MEILLEURE PRISE EN CHARGE DES MALADES : LA NÉCESSITÉ DE S'ORGANISER À L'ÉCHELON DE L'EUROPE

Chair / Modérateur : Frédéric Sicard, Health Ministry / Ministère de la Santé

European Expertise Network: State of the Art and Perspectives

Les réseaux européens de référence : état des lieux et perspectives

Alexandra Fourcade, Health Ministry / Ministère de la Santé

An European Network example

Un exemple de réseau européen

Hervé Puy, Reference centre for porphyrias / Centre de référence sur les porphyries

Pole of Competitiveness: ORPHEME

Un pôle de compétitivité : ORPHEME

Jacquie Berthe, President / Président

15.45 - 17.00 **SESSION 4**

RARE DISEASES: PATIENTS' ASSOCIATIONS, THE CORNERSTONE

LES MALADIES RARES : LE RÔLE ESSENTIEL DES ASSOCIATIONS DE MALADES

Chair / Modérateur : Dominique Donnet Kamel, INSERM

A Patients' Organisation: AFM, a Pioneering Association in the Field of Rare Diseases

Une association de malades : l'AFM, pionnière dans le domaine des maladies rares

Laurence Tiannot-Herment, President / Présidente

A National Alliance Organisation: Alliance Maladies Rares, a United Struggle for Rare Diseases and Patients

Un collectif national : l'Alliance Maladies Rares, un combat solidaire pour la reconnaissance des maladies rares et des personnes atteintes

Françoise Antonini, General Delegate / Déléguée générale

An European Umbrella Organisation: EURORDIS, a Voice for Patients with Rare Diseases in Europe

Un collectif européen : EURORDIS, une voix pour les personnes atteintes de maladies rares en Europe

Yann Le Cam, Executive Director / Directeur exécutif

17.00 **Conclusion**

Chantal Lecerf, Spokes Man Alliance des Maladies Rares / Porte Parole de l'Alliance Maladies Rares

17.15 - 17.30 BREAK / PAUSE

Friday October 10 - Vendredi 10 Octobre

17.30

ROOM THEATRE / SALLE THEATRE

OPENING CEREMONY / CEREMONY D'OUVERTURE

17.30 - 20.00

6TH WORLD RETT SYNDROME CONGRESS

OPENING CEREMONY

6ÈME CONGRÈS MONDIAL DU SYNDROME DE RETT

CÉRÉMONIE D'OUVERTURE

**LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)**

*Under the High Patronage of Mister Nicolas Sarkozy, President of the French Republic and HRH the Princess of Hanover
Organized by AFSR (French Rett Syndrome Association)*

In partnership with RSE (Rett Syndrome Europe) and IRSF (International Rett Syndrome Foundation)

*Sous le haut patronage de Monsieur Nicolas Sarkozy, Président de la République Française et de S.A.R, la Princesse de Hanovre
Organisé par l'AFSR (Association Française du Syndrome de Rett)*

En partenariat avec le RSE (Rett Syndrome Europe) et l'IRSF (Fondation Internationale sur le Syndrome de Rett)

17.30

Welcome addresses: Rare Diseases, Child Handicap, Towards Citizenship

Message de bienvenu : Les maladies rares, le polyhandicap de l'enfant, vers la citoyenneté

Didier Gaboriaud, President of FMP / Président de la FMP, La Mutualité Française

17.40

Introduction: Rare Diseases and Child handicap, Rett Syndrome a Hand Link from Gene to Care

Introduction : Maladies rares et polyhandicap de l'enfant, le Syndrome de Rett, un lien entre la génétique et les soins

Christiane Roque, President of Honour, AFSR / Présidente d'honneur, AFSR

17.50 - 18.30

OPENING LECTURES / COLLOQUES D'OUVERTURE

Chair / Modérateurs : Philippe Evrard (Paris, France), Helen Leonard (Perth, Australia)

Child Handicap, the French Model Policy

Le polyhandicap de l'enfant, vers un modèle français

Jean-François Chossy, Deputy (French Parliament) – Député (Parlement Français)

Patients' Associations Expectations to Rebuild Rituals

Les associations de Patients, entre les attentes et la reconstruction de rituels

Gérard Nguyen (Paris, France)

«Original Young Girls», a movie by AFSR

«Les jeunes filles originelles», un film de l'AFSR

Opening message from Philippe Evrard, President of the Congress

Message d'ouverture de Philippe Evrard, Président du Congrès

Welcome message from Bengt Hagberg, President of Honour of the Congress (video)

Message de bienvenue de Bengt Hagberg, Président d'honneur du Congrès (vidéo)

Opening message from Helen Leonard, President of the Programme Committee

Message d'ouverture d'Helen Leonard, Présidente du Comité de Programme

18.30 - 20.00

PLENARY SESSION / SESSION PLÉNIÈRE

Chair / Modérateurs : Masaya Segawa (Tokyo, Japan), Simone Gilgenkrantz (Nancy, France)

Inaugural Overview of Rett Syndrome, MECP2 and Beyond

Etat de l'art sur le Syndrome de Rett, MECP2 et au delà

Huda Zoghbi (Houston, USA)

MECP2, Structure and Function

MECP2, Structure et fonction

Adrian Bird (Edinburgh, UK)



A Hand Link from Gene to Care

20.30

CONGRESS GALA DINNER / DÎNER DE GALA DU CONGRÈS
SALONS DE L'HÔTEL DE VILLE DE PARIS
UNDER REGISTRATION / SUR INSCRIPTION

The Congress Reception Gala Dinner will be held on Friday at 20.30 in the reception room of the Mayor Build of Paris. This will be a fancy dress dinner on the theme of « Diversity: Culture and World Folklore ». You will find all the details in the «General Informations» page XX.

Nous avons le plaisir de vous informer que le réception dinatoire de Gala du Congrès se tiendra le Vendredi 10 octobre à 20h30 dans les salons de l'hôtel de ville de Paris. Ce sera une soirée costumée sur le thème «Diversité : Culture et Folklore du Monde». Vous trouverez toutes les informations nécessaires page XX de ce programme, au niveau des «Informations Générales».

Friday October 10 - Vendredi 10 Octobre

SATURDAY OCTOBER 11 – SAMEDI 11 OCTOBRE

**8.30 – 10.00 ROOM JUSSIEU/ SALLE JUSSIEU
PLENARY SESSION / SESSION PLENIERE**

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

Chair/ Modérateur: Philippe Evrard (Paris, France), Folker Hanefeld (Goettingen, Germany)

Clinical Overview / Etat de l'art : les aspects cliniques

Alan Percy (Birmingham, USA)

Relationship between Genotype and Phenotype / Relation Génotype / Phénotype

Helen Leonard (Perth, Australia)

**9.00 – 17.30 ROOM SAINT GERMAIN / SALLE SAINT GERMAIN
SATELITTE SYMPOSIUM / SYMPOSIUM SATELITTE**

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organized by URAPEI Ile-de-France

Journée régionale organisée par l'URAPEI Ile-de-France

**PERSONNES ATTEINTES D'AUTISME ET DE TROUBLES ENVAHISSANTS
DU DÉVELOPPEMENT (ATED) : LES FAMILLES ET LES STRUCTURES
D'ACCUEIL FACE AUX «SITUATIONS COMPLEXES».
L'ÉMERGENCE DE NOUVELLES PRISES EN CHARGE**

8.30 - 9.00 **Accueil des participants**

9.00 – 9.10 **Ouverture**

Marie-France Epagneul, Présidente de l'URAPEI Ile-de-France

9.10 – 9.25 **Articulation entre le sanitaire et le médico-social : une nécessité**

Annick Deveau, Directrice adjointe, Direction régionale des affaires sanitaires et sociales (DRASS) d'Ile-de-France

Catherine Martin-Leray, Agence régionale d'hospitalisation (ARH) (sous réserve)

9.25 – 10.30 **Actualités de la recherche sur les questions de l'autisme et des TED**

Catherine Barthelemy, pédopsychiatre, physiologiste, professeur et chef de service au CHU de Tours

9.45 – 10.00 **Débat avec la salle**

Modérateurs : Annick Deveau et Catherine Martin-Leray

10.00 - 10.30 **Pause**

10.30 - 12.00 **Regards croisés : familles, professionnels, médecins**

Marie Agnès Bouthier, parent, Seine-et-Marne

Antoine Lantier, directeur de la MAS de Domont, Val-d'Oise

Claudie Maloumbi, parent, Val-d'Oise

Sylvie Brylinski, directrice du foyer de vie « Le cèdre bleu », Hauts-de-Seine

Gérard Dubret, psychiatre, chef du Pôle de psychiatrie -CH René Dubos, Pontoise

David Cohen, pédopsychiatre, professeur à l'Université Pierre et Marie Curie et chef de service pédopsychiatrie, CHU La Pitié-Salpêtrière, Paris

11.45 - 12.00 **Débat avec la salle**

Modérateurs : Pierre Nguyen, pédopsychiatre libéral et à l'IME «La Chamade» Val-d'Oise

Hélène Rippoli, parent, Val-de-Marne



A Hand Link from Gene to Care

- 12.00 - 14.00 **Déjeuner libre**
- 14.00 - 14.45 **L'expérience d'un service spécialisé pour personnes avec autisme en Belgique**
Eric Willaye, docteur en psychologie, directeur du Service Universitaire Spécialisé pour personnes avec Autisme, (SUSA asbl) Université de Mons-Hainaut, Mons, Belgique
- 14.30 - 14.45 **Débat avec la salle**
Modérateurs : Laurence Robel, praticien hospitalier, responsable de la Consultation autisme dans le service du professeur Bernard Golse, Hôpital Necker-enfants-malades, Paris et Catherine Dupont Le Calvez (Association AIDERA-Yvelines/URAPEI)
- 14.15 - 15.30 **Unités mobiles et hospitalisation temporaire : 10 ans de préparation d'un dispositif dédié aux « Situations Complexes en ATED »**
Moïse Assouline, médecin directeur de l'Hôpital de Jour Santos-Dumont, Paris, Marie-Agnès Bouthier, parent et ex présidente de la PEPA (CRAIF) Brigitte Guiraudie, médecin inspecteur de santé publique, DASS de Paris (sous réserve)
- 15.15 - 15.30 **Débat avec la salle**
Modérateurs : Alain-Pierre Peyraud, pédopsychiatre, médecin coordinateur du pôle enfant de la MDPH de Paris et Hélène Algoud, parent, Paris
- 15.30 - 16.00 **Pause**
- 16.00 - 17.15 **L'expérience de l'ADAPEI des Deux-Sèvres (79) : collaborer avec le secteur sanitaire en faveur de l'accompagnement des adultes atteints d'autisme**
Séverine Recordon-Gavoriaud, docteur en psychologie, directrice de la Maison Pour l'Autisme, ADAPEI 79 Jean Michel Husse, cadre de santé du Centre Expertise Autisme pour Adultes du Centre Hospitalier de Niort, Deux-Sèvres
- 16.45 - 17.15 **Débat avec la salle**
Modérateurs : Jacques Piant, médecin pédopsychiatre, praticien, responsable de pôle, CH Gonesse et Elisabeth Chancerel, administratrice URAPEI, parent, Paris (sous réserve)
- 17.15 **Conclusion**
Elisabeth Gourevitch, vice-présidente du Conseil régional d'Ile-de-France, présidente du CRCCH

10.00 - 10.30 ROOM MONGE / SALLE MONGE
BREAK - VISIT OF THE POSTERS AND THE EXHIBITION
PAUSE, VISITE DES POSTERS ET DE L'EXPOSITION

10.30 - 12.00 ROOM JUSSIEU / SALLE JUSSIEU
WORKSHOP / ATELIER

LANGUAGE: FRENCH (SIMULTANEOUS TRANSLATION INTO ENGLISH AND ITALIAN)
LANGUE : FRANÇAIS (TRADUCTION SIMULTANÉE EN ANGLAIS ET EN ITALIEN)

BREATHING AND AUTONOMIC DYSFUNCTION IN RETT SYNDROME TROUBLES RESPIRATOIRES ET DU SYSTÈME NERVEUX AUTONOME

Aims:

- to demonstrate the importance of the brainstem in Rett syndrome
- to present the results obtained using the mouse models of RS to study brainstem dysfunction
- to explain what therapeutic approaches are being / could be developed to treat autonomic dysfunction in RS patients

Target Audience: Researchers, clinicians, caregivers and students

Moderator: Laurent Villard (Marseille, France)

Clinical Features of Breathing and Autonomic Abnormalities in Rett Syndrome

Peter O. O. Julu, (London, UK) – Ingegerd Witt Engerström (Frösön, Sweden)

Substrates of Respiratory and Autonomic Dysfunction in Mouse Models of Rett Syndrome

David M. Katz (Cleveland, OH, USA)

Pharmacological Treatment of Autonomic Dysfunctions in Rett Syndrome, from Mice to Men

Jean-Christophe Roux (Marseille, France)

**10.30 – 12.00 ROOM ODEON / SALLE ODEON
WORKSHOP / ATELIER**

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

**THE CLINICAL MANAGEMENT OF SCOLIOSIS IN RETT SYNDROME
LA PRISE EN CHARGE CLINIQUE DE LA SCOLIOSE DANS LE
SYNDROME DE RETT**

Aims:

- To discuss main issues that relate to the conservative strategies of monitoring of scoliosis, bracing and physiotherapy
- To discuss the indications for and outcomes of scoliosis surgery as well as the assessment and management of functional skills during the peri-operative period
- To consider a family perspective on the conservative and surgical management of scoliosis

Target Audience: Developmental paediatricians, child neurologists, neurologists, therapists, orthopaedic surgeons and researchers with an interest in scoliosis.

Moderator: *Jenny Downs (Perth, Australia)*

Introduction to Scoliosis Workshop

Jenny Downs (Perth, Australia)

Assessment, Monitoring, Bracing for Scoliosis; Surgery for Scoliosis; the Non-Surgical Management of Severe Scoliosis

David Roye (New York, USA)

Physiotherapy for Scoliosis; the Non-Surgical Management of Severe Scoliosis

Meir Lotan (Tel Aviv, Ariel, Jerusalem, Israel)

Functional Assessment and Management in the Peri-operative Period

Eva-Leena Larsson (Linköping, Sweden)

A Family Perspective on Issues of Conservative and Surgical Management

Susan L. Hall (Birmingham, UK)

**10.30 – 12.00 ROOM CONSEIL / SALLE CONSEIL
ORAL COMMUNICATIONS / COMMUNICATIONS ORALES**

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

**MOUSE MODELS 1
MODÈLES DE SOURIS 1**

Moderators: *Huda Zoghbi (Houston, USA), Walter Kaufmann (Baltimore, USA)*

Preserved Inhibitory Connectivity Patterns in the MeCP2-Stop Mutant Mouse Hippocampus

M.E.S. Bailey, S-M. Weng, S.R. Cobb (Glasgow, UK)



A Hand Link from Gene to Care

Brain Anatomy Phenotypes in Mecp2-Mutant Mouse Models of Rett Syndrome

N. Belichenko, P.V. Belichenko, H.H. Li, W.C. Mobley, U. Francke (Stanford, USA)

Dendritic and Axonal Phenotypes in MeCP2-Mutant Mouse Models of Rett Syndrome

P. Belichenko (California, USA)

Region-Specific Synaptic Abnormalities in a Mouse Model for Rett Syndrome

E. Boggio, L. Morando, M. Pallotto, M. Sassoe-Pognetto, M. Giustetto (Torino, Italy)

Detection of MeCP2 Gene Targets using Chip-on-Chip

D. Cloosterman, S. Williamson, G. Pelka, P. Tam, J. Christodoulou (Sydney, Australia)

MeCP2-Protein Substitution Therapy in a Mouse Model of Rett Syndrome

F. Laccone (Vienna, Austria)

10.30 – 12.00 ROOM POISSY / SALLE POISSY ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

GENOTYPE-PHENOTYPE

GÉNOTYPE-PHÉNOTYPE

Moderators: Helen Leonard (Perth, Australia), Carolyn Ellaway (Westmead, Australia)

The Phenotype of Rett Syndrome Cases with Deletions in the C-Terminal Region of MeCP2

A. Bebbington, H. Leonard, P. Carter, D. Ravine, N. De Klerk, J. Christodoulou (Perth, Sydney, Australia)

Rett Syndrome in Spain, Genotype-Phenotype Correlations of the Most Frequent Mutations

A. Roche, M. Pineda, A. Aracil, M. Naudó, L. Martorell, J. Armstrong (Barcelona, Spain)

Molecular Testing of MeCP2 Gene in Greek Children with MR

S. Psoni, H. Fryssira, C. Sofocleous, N. Vogiatzakis, S. Kitsiou, J. Traeger-Synodinos (Athens, Greece)

Extending the Phenotype Associated with MeCP2 Mutations in Females

P. Carter, K. Gibson, L. Nagarajan, D. Ravine, M. Davis, H. Leonard (Perth, Australia)

A New Family Case of Rett Syndrome: Two Half Sisters and One Brother with Identical MeCP2 Mutation, but with Different Clinical Presentation.

K. Ravn, GAA. Herder, JB. Nielsen, M. Schwartz, OH. Skjeldal (Glostrup, Denmark, Bodø, Oslo Norway)

A Male Rett Patient and Five Females with Preserved Speech Variant Share the Same MeCP2 Mutation

S. Russo, M. Masciadri, R. Lupi, I. Moroni, L. Angelini, L. Giordano, P. Veggiotti, F. Cogliati, MT. Bonati, L. Larizza (Milano, Brescia, Italy)

10.30 – 12.00 ROOM SORBONNE / SALLE SORBONNE SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

PSYCHOMOTRICITY

PSYCHOMOTRICITÉ

Aims / Objectifs :

We will resume in the workshop principle bases of child development and suggest techniques and exercises in correspondence with each step. In a second section, we discuss a more detailed and precise, the basement of the psychomotor; ie the sensory-motor approach/ Nous présentons dans l'atelier les principe de bases du développement de l'enfant et proposerons des techniques et

Saturday October 11 - Samedi 11 Octobre

exercices en correspondance avec chaque étape. Dans une seconde partie, nous aborderons de façon plus fine et précise, les soubassements de la psychomotricité, c'est-à-dire la sensorimotricité.

Target Audience: Parents and caregivers / Parents et professionnels

Moderators / Modérateurs: Evelyne Camaret (AFSR, France), Philippe Kostka (AFSR, France)

10.30 – 12.00 ROOM CLUNY / SALLE CLUNY
SATELLITE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

CONDUCTIVE PEDAGOGY
PÉDAGOGIE CONDUCTIVE

Aims / Objectifs :

- Overview of the theory and the principles of the Conductive Pedagogy Approach

Aperçu des principes de l'approche de la Pédagogie Conductive

- Throughout a film showing the application of the principles with a group of disabled children:

Experience sharing and discussion with parents / A travers un film montrant les applications avec un groupe d'enfants ayant un polyhandicap: partage d'expérience et discussion avec les parents

Target Audience: Caregivers working with disabled children and families / Professionnels et familles

Moderator / Modérateur: Annick Champolion-Puel (AFSR, France)

12.00 – 14.00 ROOM MONGE / SALLE MONGE
BREAK & VISIT OF THE POSTERS AND THE EXHIBITION
PAUSE, VISITE DES POSTERS ET DE L'EXPOSITION

12.00 – 13.00 ROOM JUSSIEU / SALLE JUSSIEU
BEST OF THE DAY / « BEST OF » DU JOUR

LANGUAGE: FRENCH (SIMULTANEOUS TRANSLATION INTO ITALIAN)
LANGUE : FRANÇAIS (TRADUCTION SIMULTANÉE EN ITALIEN)

AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organized by the Scientific Committee of the French Association

Organisé par le Comité Scientifique de l'AFSR

KEY NOTE ON TAKE HOME MESSAGES

ACTUALITÉS SCIENTIFIQUES DE LA JOURNÉE ET QUESTIONS / RÉPONSES

12.15 – 13.15 ROOM MONGE / SALLE MONGE
POSTERS SESSION / SESSION POSTERS

Authors will present their poster during the Posters Session:

- to present orally (5 minutes their work)

- to answer to questions

Posters will be displayed and available for independent viewing during the whole congress (October 10-13).

Posters' Thematic:

Genetics

Moderator: TBC

Clinical Investigation

Moderator: TBC

Clinical Evaluation

Moderator: TBC

Clinical Aspects

Moderator: TBC

Management

Moderator: TBC

14.00 – 15.30

ROOM JUSSIEU / SALLE JUSSIEU

PARENTS' WORKSHOP / ATELIER DE PARENTS

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

GETTING OLDER LIVING WELL

VIELLIR EN PRÉSERVANT SA QUALITÉ DE VIE

Aims:

- To highlight the problems that women with Rett syndrome face as they grow older. These include the common problems of autonomic dysfunction, increasing severity of scoliosis, increasing immobility and worsening musculoskeletal problems, which often occur in the context of poorer feeding and difficulty in maintaining weight.
- To discuss available treatment and therapy options
- To discuss some of the difficult decisions and dilemmas that families face in managing these problems

Target Audience: Parents, families and health professionals

Moderator: Hilary Cass (London, UK)

Introduction

Hilary Cass (London, UK)

A parent's journey

Yvonne Milne (Worcs, UK)

Medical interventions

Ingegerd Witt Engerström (Östersund, Sweden)

Therapy interventions

Sheena Reilly (Melbourne, Australia)

Real choices

Panel and audience

14.00 – 15.30

ROOM ODEON / SALLE ODEON

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

PHENOTYPE AND FUNCTION

PHÉNOTYPE ET FONCTION

Moderators / Modérateurs : Alan Percy (Birmingham, USA), Walter Kaufmann (Baltimore, USA)

Is there an Early Specific Developmental Trajectory of Rett Disorder?

C. Einspieler, P.B. Marschik, H.F.R. Prechtl, A.M. Kerr, F. Ferrari, M.F. Roversi, G. Cioni (Graz, Austria - Glasgow, UK – Modena, Pisa, Italy)

Gross Motor Skills of 99 Females with Rett Syndrome

J. Downs, A. Bebbington, P. Jacoby, H. Leonard (Perth, Australia)

General Development in Females with Rett Syndrome, Focusing on Abilities, Deformities and Management: The Swedish Rett Center Survey

E. Larsson, B. Lindström, I. Witt Engerström (Umeå, Frösön, Sweden)

Autism and Rett Syndrome - How Different the Differences are?

A. Hill (Gosford, Australia)

Rett Syndrome and Rett Disorder: An Attempt to Redefine Phenotypes

T. Temudo, M. Santos, E. Ramos, K. Dias, JP. Vieira, A. Moreira, E. Calado, I. Carrilho, J. Sequeiro, P. Maciel (Porto, Braga, Portugal)

Diagnostic Criteria for the Zappella Variant of Rett Syndrome (The Preserved Speech Variant)

M. Zappella, F. Mari, MA. Mencarelli, E. Scala, F. Ariani, I. Longo, I. Meloni, G. Pini, G. Hayek, A. Renieri (Viareggio, Siena Italy)

ROOM CONSEIL / SALLE CONSEIL

14.00 – 15.30 WORKSHOP / ATELIER

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

MOLECULAR PATHOGENESIS OF RETT SYNDROME

PATHOPHYSIOLOGIE MOLÉCULAIRE ET SYNDROME DE RETT

Aims:

- To discuss experiments designed to understand the role of MeCP2 in DNA-methylation dependent chromatin regulation
- To discuss the use of genetically modified mouse models in investigating the molecular pathogenesis of Rett syndrome
- To compare/contrast MeCP2 and MBDs' role in gene regulation in cancer versus Rett syndrome

Target Audience: Scientists, physicians

Moderator / Modérateur : Janine LaSalle (Davis, USA)

MeCP2 and DNA Methylation Targets in Rett Syndrome and Cancer Models

Esteban Ballestar (Barcelona, Spain)

MeCP2-Dependent Transcriptional Repression is Required for Behavioral Modalities Mediated by the Basolateral Amygdala

Megumi Adachi (Dallas, USA)

Neuron-Specific Expression of Setdb1 Histone Methyltransferase in a Mouse Model of Rett Syndrome

Schahram Akbarian (Worcester, USA)

ROOM POISSY / SALLE POISSY

14.00 – 15.30 ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

MANAGEMENT OF EPILEPSY AND CARDIORESPIRATORY CHALLENGES IN RETT SYNDROME

PRISE EN CHARGE DE L'ÉPILEPSIE ET DES DÉSORDRES CARDIORESPIRATOIRES DANS LE SYNDROME DE RETT

Moderators / Modérateurs : Nadia Bahi-Buisson (Paris, France), Bruria Ben Zeev (Ramat-Gan, Israel),

A Hand Link from Gene to Care

EEG Characteristics of Rett Syndrome

X. Bao, H. Lu, G. Cao, X. Liu, H. Pan, X. Wu (Beijing, China)

Epilepsy in Rett Syndrome: A Study of Association between Phenotype and Genotype

B. Cardoza, M. Kerr, A. Clarke, F. Gibbon, P. Smith, J. Wilcox (Cardiff, UK)

Epilepsy in Rett Syndrome

M. Djuric, D. Zamurovic, R. Kravljanac, G. Vlahovic, B. Vucetic (Belgrade, Serbia)

The Common BDNF Polymorphism may be a Modifier of Disease Severity in Rett Syndrome

B. Ghidoni Ben-Zeev, A. Bebbington, G. Ho, H. Leonard, N. De Klerk, E. Gak, M. Veckler, J. Christodoulou (Ramat-Gan, Tel-Aviv, Israel – Sydney, Australia)

Epilepsy in Rett Syndrome

M. Pintaudi, A. Vignoli, F. Labriola, MG. Baglietto, L. Giordano, J. Galli, E. Veneselli, MP. Canevini (Genoa, Milan, Brescia, Italy)

Clinical Update Addressing the Cardiorespiratory Challenges in Medicine Posed by Rett Syndrome: The Frösö Declaration

E. Smeets, P.O. Julu, I. Witt-Engerstrom, S. Hansen, F. Apartopopoulos, B. Wiit (The Netherlands - London, Glasgow, Uk – Östersund, Sweden – Viareggio, Italy)

Oxidative Stress in Rett Syndrome

J. Hayek, C. De Felice, C. Signorini, S. Leoncini, M. Rossi, G. Latini, M. Comporti, L. Ciccoli (Siena, Lecce, Italy)

ROOM SORBONNE / SALLE SORBONNE

14.00 – 15.30 ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

FAMILY ISSUES & EXPERIENCES

EXPÉRIENCES ET ATTENTES DES FAMILLES

Moderators / Modérateurs : Pascaline Guerin (Chartres, France), Gérard Nguyen (Paris, France),

Communication as a Basis and Means for Interaction, Play and Learning for Girls with Rett Syndrome

E. Saraf, B. Ben-Zeev, J. Wine (Ramat-Gan, Israel)

Limitations or Possibilities – A Case Study

H. Andersen, G. Roende, J.B. Nielsen, S. Blichfeldt (Glostrup, Denmark)

Living with Rett Syndrome: Family Stories of Coherence and Resilience

R. Retzlaff (Heidelberg, Germany)

The Quality of Life in the Girls with Rett Syndrome

M. Roccella, L. Parisi, T. Di Filippo, D. Testa (Palermo, Italy)

Rett Syndrome – To Teach Me is to Understand Me

E. Saraf (Ramat-Gan Israel)

The Experiences of Parents who have a Child Diagnosed with a Genetic Syndrome: Rett Syndrome

H. Turner, K. Featherstone (Cardiff, Uk)

ROOM CLUNY / SALLE CLUNY

14.00 – 15.30 SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

DAY TO DAY MANAGEMENT, IMPLEMENTING BASAL STIMULATION (DESCRIBED BY ANDREAS FRÖHLICH)

Saturday October 11 – Samedi 11 Octobre

L'APPROCHE AU QUOTIDIEN DES PERSONNES AYANT UN POLYHANDICAP, EN RÉFÉRENCE À L'APPROCHE DE LA STIMULATION BASALE* D'ANDREAS FRÖHLICH

Aims / Objectifs :

Introduction on the concept and basic principles of this approach, leaders propose advice and practical ideas useful in daily support.

The experience of professionals will be complemented and enriched by a mother experience with a daughter suffering from Rett syndrome.

- Introduction on the concept and basic principles of basal stimulation approach described by Andreas Frohlich / Introduction à l'approche par la stimulation basale, méthode écrite par Andreas Frohlich*
- Implementation of daily support / Apport dans la vie au quotidien*
- Throughout a family experience with a girl with Rett Syndrome, discussion and experience sharing / A travers l'expérience d'une maman avec une fille ayant un Syndrome de Rett, discussion et partage d'expériences.*

Target Audience: Caregivers working with disabled children and families / Professionnels et familles

Moderator / Modérateur: Dany Guerlach (AFSR, France)

16.00 – 17.30 ROOM JUSSIEU / SALLE JUSSIEU **PLENARY SESSION / SESSION PLENIERE**

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

Chair/Modérateur: Mickael Johnston (Baltimore, USA)

What is MeCP2 Really Doing in the Brain?

Comment le MeCP2 agit-il réellement au niveau du cerveau ?

Janine LaSalle (Davis, CA, USA)

16.00 – 17.30 ROOM ODEON / SALLE ODEON **SATELLITE WORKSHOP / ATELIER SATELLITE**

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

DAY TO DAY STRESS MANAGEMENT **GESTION DU STRESS AU QUOTIDIEN**

Aims / Objectifs :

- Overview of stress manifestations and impact in Professional situations / Aperçu des manifestations et des conséquences du stress*
- Identification of stressful situations / Identification des situations générant du stress*
- Implementation of stress release techniques in day to day practice / Exercices pratiques de gestion du stress applicables en situation professionnelle*

Target Audience : Caregivers working with disabled children and families / Professionnels et familles

Moderator / Modérateur : Pierre Mardycs, physiotherapist, specialised in training of stress management

16.00 – 17.30 ROOM CONSEIL / SALLE CONSEIL **SATELLITE WORKSHOP / ATELIER SATELLITE**

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

CIT* AND SOCIAL CHANGES "TODAY I HAVE A DREAM ..."

***CIT = COMMUNICATION AND INFORMATION TECHNOLOGIES**



A Hand Link from Gene to Care

Aims:

- Help audience to understand why and how society is changing regarding rare diseases because of CIT,
- Explain how Rett Sd patients and relatives should use CIT to change society,
- Describe to the audience how CIT should be used for better social life.

Target Audience: Rett Sd patients' relatives and carers (personal and professional), CIT' professionals working in e-health and social networking.

Moderator: Christian Schoen, general practitioner working in CIT domain related to health ("e-health")

16.00 – 17.30

ROOM POISSY / SALLE POISSY

SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

EYE MOTRICITY

OCULOMOTRICITÉ

Aims / Objectifs:

- Anatomical and physiological of the vision overview / Rappel anatomique et physiologique de la vision
- Visual data processing overview / Rappel sur le traitement de l'information visuelle
- Focus on calibration and the hierarchical organization of the visual stimuli / Parler de la calibration et la hiérarchisation des stimuli visuels
- Discussion based on clinical cases and expectations from parents / Discussion à partir des expériences et témoignages pour répondre aux attentes des parents.

Target Audience: Caregivers working with disabled children and families / Professionnels et familles

Moderator / Modérateur : Nadia Kebbal (AFSR, France)

16.00 – 17.30

ROOM SORBONNE / SALLE SORBONNE

SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

RELATIONAL AND CLOWN GAMES

«JEU RELATIONNEL ET JEU DE CLOWN»

Aims / Objectifs:

- Find another way to play in the relationship with the other / Découvrez une autre façon de jouer dans la relation avec l'autre
- Communicate in the game in imitation, through the eyes / Communiquer par le jeu dans l'imitation, par le regard
- Playing with his emotions through the mask of red nose which authorizes and reveals the game in his poetry and its symbolism / Jouer avec ses émotions grâce au masque du nez rouge qui autorise et révèle le jeu dans sa poésie et sa symbolique .

Target Audience: Parents and caregivers / Parents et professionnels

Moderator / Modérateur : Pascale Gondebeaud-Sylvander

Specializing in communication with the work of clown theatre

Spécialisée en communication par le travail du clown de théâtre

Saturday October 11 – Samedi 11 Octobre

16.00 – 17.30 ROOM CLUNY / SALLE CLUNY
SATELLITE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

THE WORD THAT HEALS
LA PAROLE QUI GUÉRIT

A workshop discovered around the practice of storytelling
Un atelier découverte autour de la pratique du conte

Aims / Objectifs :

- Knowing “storytelling” our history / Savoir “Ra Conter” son parcours de Vie
- Since the dawn of time and in all cultures, storytelling is involved in caring for the soul. It transmits beyond stories below and in a language, laws of the World subtle. Its values are soothing intuitively understood by the listener, because they are such vibration. Storytelling could be learned, but everyone can learn the rudiments of this art of the relationship, because it is less a technique that is necessary to state that this practice inside / Depuis l’aube des temps et dans toutes les cultures, le conte participe au soin de l’âme. Il transmet, au-delà des histoires et dans un infra langage, les lois du monde subtil. Ses valeurs apaisantes sont comprises intuitivement par celui qui écoute, car elles sont de nature vibratoire. Conter s’apprend, certes, mais tout le monde peut apprendre les rudiments de cet art de la relation, car c’est moins une technique qui est nécessaire à cette pratique qu’un état intérieur
- It is around this inner state that we will work in this workshop / C’est autour de cet état intérieur que nous travaillerons dans cet atelier.

Target Audience: Parents and caregivers / Parents et professionnels

Moderator / Modérateur : Martine Tollet

Storyteller and art therapist

Conteuse et art thérapeute

17.30 – 19.30 ROOM JUSSIEU / SALLE JUSSIEU
SATELLITE SYMPOSIUM / SYMPOSIUM SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

AUDIENCE: CLINIENS, PROFESSIONNELS DE SANTÉ, ASSOCIATIONS DE PATIENTS

Organized by SFNP (French Society of Neuropaediatrics)

Organisé par la SFNP (Société Française de Neuropédiatrie)

ENJEU ET PERSPECTIVE DES ESSAIS THÉRAPEUTIQUES DANS LE SYNDROME DE RETT

Modérateur : Philippe Evrard (Paris, France)

Essais thérapeutiques chez l’enfant ; principe des essais et problématique des petites populations et des maladies rares

Catherine Chiron, INSERM U663 Paris Descartes

Desipramine dans les troubles ventilatoires du syndrome de Rett ;

Essais en double aveugle contre placebo

Josette Mancini, Neuropédiatrie Hopital La Timone Marseille

Fluoxétine dans le syndrome de Rett ; un modèle d’essai translationnel et principe de l’essai ouvert

Nadia Bahi-Buisson, Neuropédiatrie Hôpital Necker Enfants Malades APHP Université Paris Descartes

Discussion et Perspectives

17.30 – 19.30

ROOM ODEON / SALLE ODEON

SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

CARDIORESPIRATORY DYSFUNCTION AND MANAGEMENT
IN RETT SYNDROME

TROUBLES CARDIORESPIRATOIRES ET PRISE EN CHARGE DANS LE
SYNDROME DE RETT

Aims:

- To learn about three distinct cardiorespiratory phenotypes in Rett Syndrome
- To learn the clinical risks unique to each of the three cardiorespiratory phenotypes in Rett Syndrome
- To learn of various clinical interventions available for each of the three cardiorespiratory phenotypes in Rett Syndrome

Target Audience: physicians, caregivers and parents

Moderator / Modérateur : Peter O. O. Julu (London, UK), Ingegerd Witt Engerström (Östersund, Sweden)

Clinical Determination of Cardiorespiratory Phenotypes in Rett Syndrome

Peter O. O. Julu (London, UK)

Cardiorespiratory Characterisation in Rett Syndrome: The Italian Experience

Giorgio Pini (Lido di Camaiore, Italy)

Significance of Cardiorespiratory Phenotypes in Clinical Management of Rett Syndrome

Peter O. O. Julu (London, UK)

Cardiorespiratory Influences on the EEG in Rett Syndrome

Stig Hansen (Glasgow, UK)

Abnormal Spontaneous Brainstem Activities (ASBA) in Rett Syndrome

Peter O. O. Julu (London, UK)

Treatment of Apneustic Breathing in Girls with Rett Syndrome- A Pilot Study

Sami S.F. Al-Rawas (Muscat, Oman)

Detection of Hypocapnic Attacks and Resuscitation in Rett Syndrome

Stig Hansen (Glasgow, UK)

SUDEP: Sudden Unexpected Death in Epilepsy: Lessons from Rett Syndrome

Robert S. Delamont (London, UK)

The Use of Cardiorespiratory Indices in the Management of Patients with Rett Syndrome

Ingegerd Witt Engerström (Östersund, Sweden)

17.30 – 19.00

ROOM POISSY / SALLE POISSY

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE: ANGLAIS

MOUSE MODELS 2

MODÈLE DE SOURIS 2

Chairs / Modérateurs : Janine LaSalle (Davis, USA), David Ravine (Perth, Australia)

Expression of Methyl CPG Binding Protein 2 (MeCP2) during the Postnatal Development of the Mouse Brainstem

E. Dura, L. Villard, J.C. Roux (Marseille, France)

The MeCP2-Null Mouse Hippocampus is Prone to Hyper-Excitability and Displays an Altered Basal Inhibitory Rhythm

J. Eubanks, L. Zhang (Toronto, Canada)

Cortical and Hippocampal EEG Recordings from MeCP2-Deficient Mice Reveal Alterations in Basal Brain Wave Activity, and Heightened Sensitivity to Agonist-Induced Epileptiform Discharge

J. Eubanks, J. D'Cruz, C. Wu, L. Zhang (Toronto, Canada)

Introduction of Heterozygous Reln Deletion Results in Extended Lifespan and Delayed Onset of Cognitive Deficit in MeCP2-Deficient Male Mice

U. Francke, HH. Li, C. Jordan, F. Ding (Stanford, Usa)

MeCP2 Directly Regulates Insulin-Like Growth Factor Binding Protein 3 Expression in Brains

M. Itoh (Kodaira JAPAN)

Prevention of Cortical Synaptic Alterations and Improvement of Behavioural Deficits of Male and Female MeCP2 Null Mice by Early Environmental Stimulation

G. Lonetti, A. Angelucci, E. Boggio, L. Morando, M. Giustetto, T. Pizzorusso (Pisa, Florence, Turin, Italy)

ROOM SORBONNE / SALLE SORBONNE

17.30 – 19.30 BEST OF THE DAY / « BEST OF » DU JOUR

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organized by the Scientific Committee of the French Association

Organisé par le Comité Scientifique de l'AFSR

KEY NOTE ON TAKE HOME MESSAGES

ACTUALITÉS SCIENTIFIQUES DE LA JOURNÉE ET QUESTIONS / RÉPONSES

ROOM SAINT GERMAIN / SALLE SAINT GERMAIN

17.30 – 19.30 SATELLITE SYMPOSIUM / SYMPOSIUM SATELLITE

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

AUDIENCE: PARENTS ASSOCIATIONS AND RESEARCHERS / ASSOCIATIONS DE PARENTS ET CHERCHEURS

Organized by EWGRS (European Working Group of Rett Syndrome)

Organisé par EWGRS (Groupe de travail Européen sur le Syndrome de Rett)

EUROPEAN WORKING GROUP ON RETT SYNDROME (EWGRS)

Chair: Charlotte Kilstrup-Nielsen (Busto Arsizio, Italy)

17.30-17.50

The European Working Group on Rett Syndrome (EWGRS) in its First Year: Objectives and Accomplishments

Nicoletta Landsberger (Busto Arsizio, Italy)

17.50-18.20

Eurorett the First Spin-Off of the EWGRS: Where we are and where are we going

Laurent Villard (Marseille, France)

18.20-18.50

Rett Syndrome Needs a Therapy; A Proposal to Accelerate its Finding

Prorrett Ricerca

19.19-19.0

Discussion

SUNDAY OCTOBER 12 – DIMANCHE 12 OCTOBRE

Sunday October 12 – Dimanche 12 Octobre

8.00 – 10.00

ROOM JUSSIEU / SALLE JUSSIEU

PLENARY SESSION / SESSION PLENIERE

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

OTHER PHENOTYPES AUTRES PHÉNOTYPES

Chair: Sakkubai Naidu (Baltimore, USA)

Male Phenotype / Autres Phénotypes

Hilde Van Esch (Leuven, Belgium)

Congenital Rett Syndrome Variant / Variante congénitale du Syndrome de Rett

Alessandra Renieri (Siena, Italy)

CDKL5

John Christodoulou (Westmead, Australia)

Rett Syndrome and Autism: What is the Relationship? / Syndrome de Rett et autisme : quel est le lien ?

Walter Kaufmann (Baltimore, USA)

10.00 – 10.30

ROOM MONGE / SALLE MONGE

BREAK - VISIT OF THE POSTERS AND THE EXHIBITION
PAUSE, VISTE DES POSTERS ET DE L'EXPOSITION

10.30 – 12.00

ROOM JUSSIEU / SALLE JUSSIEU

WORKSHOP / ATELIER

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

GASTROINTESTINAL (FEEDING, SWALLOWING AND DIGESTIVE ABILITY) PROBLÈMES DIGESTIFS ET TROUBLES ALIMENTAIRES

Aims:

- Outline the evidence around problems associated with swallowing and feeding in Rett syndrome
- Provide a parent's perspective of the issues and concerns around feeding and swallowing in Rett syndrome
- Discuss issues of management including
 - What are the best ways to monitor feeding and swallowing problems?
 - Are worsening swallowing problems/dysphagia predicatable?
 - Advantages and disadvantages of gastrostomy
 - Is fundoplication a good operation for girls with Rett syndrome?

Target Audience: Researchers, clinicians and families

Chair: Susan Fyfe (Perth, Australia)

DATA BASED DISCUSSION OF ISSUES

- **SOMA – Oral dysphagia**
Gordon Baikie (Melbourne, Australia)
- **Feeding impairment and Hypergag**
Catherine Senez (Toulouse, France)

- Parent perspective of feeding problems in Rett syndrome

Kathy Hunter (IRSF, USA)

- BMI feeding problems

Susan Fyfe (Perth, Australia)

MANAGEMENT OF FEEDING PROBLEMS

- Aspiration, constipation

Gordon Baikie (Melbourne, Australia)

- Parents views on gastrostomy findings from our InterRett study

Susan Fyfe (Perth, Australia)

PANEL DISCUSSION

ROOM ODEON / SALLE ODEON

10.30 – 12.00 ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

NATURAL HISTORY

HISTOIRE NATURELLE DE LA MALADIE

Chairs / Moderateurs : Helen Leonard (Perth, Australia), Alan Percy (Birmingham, USA)

Movement Disorders in Rett Syndrome: An Analysis of 60 Patients with Detected MeCP2 Mutation and Correlation with Mutation Type

T. Temudo, E. Ramos, K. Dias, C. Barbot, J.P. Vieira, A. Moreira, E. Calado, I. Carrilho, J. Sequeiros, P. Maciel (Porto, Braga, Portugal)

Evolution of Stereotypies in Adolescents and Women with Rett Syndrome

A. Vignoli, F. La Briola, M.P. Canevini (Milan, Italy)

Rett Syndrome in Adult Women: Data from the British Isles Rett Syndrome Survey (Bis)

A. Hryniewiecka-Jaworska, H. Archer, A. Clarke (Cardiff, UK)

Survival Rate of the Originally Described Females with the Rett Syndrome.

M. Freilinger, I. Lanator, D. Dunkler, G. Bernert, GM. Ronen, R. Seidl (Vienna, Austria - Ontario Canada)

Aging in People With Specific Genetic Syndromes: The Rett Syndrome

S. Smeets Eric, N. Halbach, C. Schrandner-Stumpel, M. Maaskant, H. Lantman-De Valk, L. Curfs (Maastricht, The Netherlands)

The Relationship between Morbidity and Health Service Use over Time in a Rett Syndrome Population

A. Bebbington, D. Young, N. Deklerk, C. Bower, L. Naragajan, H. Leonard (Perth, Australia)

ROOM CONSEIL / SALLE CONSEIL

10.30 – 12.00 WORKSHOP / ATELIER

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

COGNITION AND COMMUNICATION

COMPRÉHENSION ET COMMUNICATION

Aims:

- Clear understanding of the language abilities that girls with Rett syndrome have and can develop
- Applying state of the art alternative technology to help individuals with Rett Syndrome access and interact with their environment
- Educational interventions which have successfully demonstrated the cognitive abilities of girls with Rett syndrome



A Hand Link from Gene to Care

Target Audience: Physicians and subspecialists, psychologists, speech language pathologists, augmentative communication specialists, therapists, educators, parents

Chair: Sarojini S. Budden (Portland, USA)

Augmentative and Alternative Communication Intervention in Girls with Rett Syndrome
Judy Wine (Jerusalem, Israel)

Assessing and Improving Communication Skills-Some Exciting Advances
Nigel Livingston (Victoria, BC, Canada)

Measures and Empowerment of Cognitive Abilities in Rett Syndrome
Rosa Angela Fabio (Milano, Italy), Sarojini S. Budden (Portland, USA)

10.30 – 12.00 ROOM POISSY / SALLE POISSY ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

CLINICAL AND THERAPEUTIC MANAGEMENT PRISE EN CHARGE CLINIQUE ET THÉRAPEUTIQUE

Moderators / Modérateurs : Jenny Downs (Perth, Australia), Luciana Scattin (Paris, France)

Clinical Intervention; Regaining or Retraining Function and Examples of Motivating Activities
E. Larsson (Umeå, Sweden)

Long-Term Follow-up of Functioning after Spinal Surgery in Patients with Rett Syndrome
E. Larsson, S. Aaro, P. Ahlinder, H. Normelli, H. Tropp, B. Öberg (Linköping, Sweden)

Surgery for Scoliosis - and Afterwards. Case Reports
E. Larsson (Frösön, Umeå, Sweden)

Improving Functional Skills and Physical Fitness in Children with Rett Syndrome
M. Lotan (Ariel, Ramat Gan, Israel)

Postoperative Care after Vertebral Arthrodesis of Children with Rett Syndrome.
H. Ben Ameer, A. A. Dalhoumi, M. Bouali, H. Amthor, R. Rubinsztajn, J. Bataille (Garches, France)

The Effectiveness of Light-Weight Dynamic Thermoplastic Hand Splints in Reducing Stereotypical Hand-Movements in 3 Girls with Rett Syndrome
A. Blore (Tadworth, UK)

10.30 – 12.00 ROOM SORBONNE / SALLE SORBONNE SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

RELATIONAL AND CLOWN GAMES «JEU RELATIONNEL ET JEU DE CLOWN»

Aims / Objectifs:

- Find another way to play in the relationship with the other / Découvrez une autre façon de jouer dans la relation avec l'autre
- Communicate in the game in imitation, through the eyes / Communiquer par le jeu dans l'imitation, par le regard
- Playing with his emotions through the mask of red nose which authorizes and reveals the game in his poetry and its symbolism / Jouer avec ses émotions grâce au masque du nez rouge qui autorise et révèle le jeu dans sa poésie et sa symbolique «.

Sunday October 12 – Dimanche 12 Octobre

Target Audience: Parents and Caregivers / Parents et Professionnels

Moderator / Modérateur : Pascale Gondebeaud-Sylvander

Specializing in communication with the work of clown theatre

Spécialisée en communication par le travail du clown de théâtre

10.30 – 12.00 ROOM CLUNY / SALLE CLUNY
SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

ACCOMPANYING THE CRIES

ACCOMPAGNER LES CRIS

Aims / Objectifs :

- *As the noises that sometimes beset us, cries can be difficult to bear / Comme les bruits qui parfois nous assaillent, les cris peuvent être difficiles à supporter.*
- *Can we come into resonance with them? / Peut-on entrer en résonance avec eux ?*
- *Can we, with our body, our voice, hear and respond, as we would with a language, or music? / Peut-on, avec notre corps, notre voix, les entendre et leur répondre, comme on le ferait avec un langage, ou une musique ?*
- *We will all experience during the workshop / Nous en ferons ensemble l'expérience au cours de l'atelier.*

Target Audience: Parents and Caregivers / Parents et Professionnels

Moderator / Modérateur : Emmanuelle Parrenin

Artist musician - Specializing in vocal work

Artiste musicienne - Spécialiste en travail vocal

12.00 – 13.00 ROOM JUSSIEU / SALLE JUSSIEU
BEST OF THE DAY / « BEST OF » DU JOUR

LANGUAGE: FRENCH (SIMULTANEOUS TRANSLATION INTO ITALIAN)
LANGUE : FRANÇAIS (TRADUCTION SIMULTANÉE EN ITALIEN)

AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

KEY NOTE ON TAKE HOME MESSAGES

ACTUALITÉS SCIENTIFIQUES DE LA JOURNÉE ET QUESTIONS / RÉPONSES

14.00 – 15.30 ROOM JUSSIEU / SALLE JUSSIEU
WORKSHOP / ATELIER

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

OUTCOME MEASURES FOR CLINICAL TRIALS IN RETT SYNDROME
RÉSULTAT DES ESSAIS CLINIQUES DANS LE SYNDROME DE RETT

Aims:

- *Outline outcome measures which would be sensitive to changes in functional ability for use in clinical trials in Rett syndrome*
 - *Discuss issues of developing measures specific for clinical trials*
 - *Hand use in Rett syndrome as a measure of functional ability*

A Hand Link from Gene to Care

- Developing statistically-based measurement of severity using existing questionnaire data
- Review the most useful measures used in studies in Rett syndrome

Target Audience: Researchers, clinicians, therapists

Chair: Sue Fyfe (Perth, Australia)

DATA BASED DISCUSSION OF ISSUES

Outcomes measures in clinical trials- the most useful measures

Sakkubai Naidu (Baltimore, USA)

Hand use in Rett syndrome as a measure of functional ability

Jenny Downs (Perth, Australia)

Developing statistically-based measurement of severity using existing questionnaire data

Ami Bebbington (Perth, Australia)

Development of outcome measures Panel discussion and questions

Helen Leonard, Carolyn Schanen, Walter Kaufmann, Alan Percy, Omar Kwaja, Hayley Archer

14.00 – 15.30

ROOM ODEON / SALLE ODEON

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

CDKL5

Chairs / Modérateurs : John Christodoulou (Westmead, Australia), Silvia Russo (Cusano Milanino, Italy)

The CDKL5 Disorder - A Description of 19 Pathogenic Mutations and Associated Phenotype

H. Archer, J. Evans, K. Ravn, A. Nemeth, J. Hurst, B. Kerr, E. Mccann, R. Appleton, L. Lazarou, A. Clarke (Cardiff, Oxford, Manchester, Liverpool, UK – Copenhagen, Denmark)

Mutation Analysis of the X-Linked Cyclin-Dependent Kinase-Like 5 Gene (CDKL5) in Patients with Early-Onset Epileptic Encephalopathy and Rett-Like Phenotype.

R. Polli, S. Sartori, E. Bettella, I. Toldo, C. Boniver, M. Vecchi, L. Giordano, B. Dalla Bernardina, G. Perilongo, A. Murgia (Padua, Brescia, Verona, Italy)

CDKL5 Mutations in Rett Patients with Early Onset Epilepsy

M. Marchi, M. Pintaudi, M. Masciadri, R. Lupi, V. Saletti, E. Veneselli, B. Ben Zeev, F. Cogliati, L. Larizza, S. Russo (Milano, Genova, Italy - Ramat-Gan, Israel)

Early Onset Seizure Variant of Rett Syndrome: Toward the Definition of the Clinical Diagnostic Criteria

M. Mencarelli, R. Artuso, F. Ariani, M. Pollazzon, F. Vigevano, A. Parmeggiani, S. Buoni, J. Hayek, F. Mari, A. Renieri (Siena, Rome, Bologna, Italy)

A Novel CDKL5 Mutation in a 47, XXY Boy with the Early-Onset Seizure Variant of Rett Syndrome

G. Di Rosa, R. Polli, S. Sartori, E. Bettella, G. Tricomi, G. Tortorella, A. Murgia (Messina, Padua, Italy)

MeCP2 Regulates CDKL5 Expression by Binding to its Methylated Gene Promoter

J. Zwiller, D. Carouge, P. Anglard (Strasbourg, France)

14.00 – 15.30

ROOM CONSEIL / SALLE CONSEIL

WORKSHOP / ATELIER

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

GENE THERAPY IN MOUSE MODELS

THÉRAPIE GÉNIQUE CHEZ LES MODÈLES DE SOURIS

Aims:

- To review recent advances in the development of mouse models of Rett syndrome.
- To review different approaches for characterizing Rett-like phenotype in mice.
- To review new genetic and molecular strategies for reversing the Rett-like phenotype in mouse models.

Sunday October 12 – Dimanche 12 Octobre

Target Audience : Neuroscientists, geneticists, clinicians in general.

Chairs: James Eubanks (Toronto, Canada)

Ectopic reintroduction of MeCP2 into forebrain neurons of heterozygous Mecp2-deficient female mice improves their Rett-like phenotype

James Eubanks (Toronto, Canada)

Deficits in synapse maturation and their therapeutic reversal in a mouse model of Rett Syndrome

Mriganka Sur (Cambridge, U.S.A.)

**14.00 – 15.30 ROOM POISSY / SALLE POISSY
WORKSHOP / ATELIER**

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

**SLEEP AND BEHAVIOUR DIFFICULTIES IN RETT
ASPECTS ÉMOTIONNELS ET COMPORTEMENTAUX ET PRISE EN
CHARGE DES TROUBLES DU SOMMEIL**

Aims:

- To describe the common difficulties in these areas of function
- To review what is known of the contributory factors
- To discuss the available & potential management of these difficulties

Target Audience: Professionals and parents

Chair: Alison Kerr (Glasgow, Scotland)

Introductory Remarks

Alison Kerr (Glasgow, Scotland)

Emotional and Behavioural Aspects of Rett Syndrome: Clinical Update

Pascaline Guerin (Chartres, France)

Emotion & Behaviour in RS

Sarajini S. Budden (Portland, USA)

Sleep & Behavioural Disturbance in Rett Syndrome

Yoshiko Nomura (Tokyo, Japan)

Panel & audience discussion

**14.00 – 15.30 ROOM SORBONNE / SALLE SORBONNE
SATELLITE WORKSHOP / ATELIER SATELLITE**

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

Organized by CPME (Paramedical and Educational Committee) AFSR

Organisé par le CPME (Comité Paramédical et Educatif) AFSR

**FOOD AND NUTRITION: EVALUATION AND MANAGEMENT IN RETT
MANAGEMENT**

**ALIMENTATION, NUTRITION ET SYNDROME DE RETT :
ÉVALUATION ET PRISE EN CHARGE**

Aims:

- Outline nutrition status assessment / *Approche de l'état nutritionnel*
- In the light of the French survey conducted within AFSR, overview of problems encountered and families' perception / *à la lumière de l'enquête faite par l'AFSR auprès des familles, revue des problèmes et de leurs expressions*



A Hand Link from Gene to Care

- To discuss on solutions and educative framework / *Discussion sur les solutions et les mesures éducatives*

Target Audience: Professionals and parents / Professionnels et parents

Chair: Irène Benigni (AFSR, France), Catherine Senez (AFSR, France)

14.00 – 15.30 ROOM CLUNY / SALLE CLUNY SATELITTE WORKSHOP ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

THE WORD THAT HEALS: A WORKSHOP DISCOVERED AROUND THE PRACTICE OF STORYTELLING

LA PAROLE QUI GUÉRIT : UN ATELIER DÉCOUVERTE AUTOUR DE LA PRATIQUE DU CONTE

Aims / Objectifs :

- Knowing “storytelling” our history / *Savoir “Ra Conter” son parcours de Vie*
- Since the dawn of time and in all cultures, storytelling is involved in caring for the soul. It transmits beyond stories below and in a language, laws of the World subtle. Its values are soothing intuitively understood by the listener, because they are such vibration. Storytelling could be learned, but everyone can learn the rudiments of this art of the relationship, because it is less a technique that is necessary to state that this practice inside / *Depuis l’aube des temps et dans toutes les cultures, le conte participe au soin de l’âme. Il transmet, au-delà des histoires et dans un infra langage, les lois du monde subtil. Ses valeurs apaisantes sont comprises intuitivement par celui qui écoute, car elles sont de nature vibratoire. Conter s’apprend, certes, mais tout le monde peut apprendre les rudiments de cet art de la relation, car c’est moins une technique qui est nécessaire à cette pratique qu’un état intérieur*
- It is around this inner state that we will work in this workshop / *C’est autour de cet état intérieur que nous travaillerons dans cet atelier.*

Target Audience: Parents and Caregivers / Parents et Professionnels

Moderator / Modérateur : Martine Tollet

Storyteller and art therapist

Conteuse et art thérapeute

16.00 – 17.30 ROOM JUSSIEU / SALLE JUSSIEU PLENARY SESSION / SESSION PLENIERE

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

INTERNATIONAL COLLABORATIONS: SETTING THE SCENE
COLLABORATIONS INTERNATIONALES : MISE EN SCÈNE

Chair / Modérateur : Helen Leonard (Perth, Australia)

InterRett

Alison May Anderson (Perth, Australia)

Rettbase

John Christodoulou (Westmead, Australia)

EuroRett

Laurent Villard (Marseille, France)

Sunday October 12 – Dimanche 12 Octobre

Rett Search

Walter Kaufmann (Baltimore, USA)

Italian Rett Biobank

Francesca Mari (Siena, Italy)

16.00 - 17.30

ROOM SORBONNE / SALLE SORBONNE

SATELITTE WORKSHOP / ATELIER SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

ACCOMPANYING THE CRIES

ACCOMPAGNER LES CRIS

Aims / Objectifs :

- *As the noises that sometimes beset us, cries can be difficult to bear / Comme les bruits qui parfois nous assaillent, les cris peuvent être difficiles à supporter.*
- *Can we come into resonance with them? / Peut-on entrer en résonance avec eux ?*
- *Can we, with our body, our voice, hear and respond, as we would with a language, or music? Peut-on, avec notre corps, notre voix, les entendre et leur répondre, comme on le ferait avec un langage, ou une musique ?*
- *We will all experience during the workshop / Nous en ferons ensemble l'expérience au cours de l'atelier.*

Target Audience: Parents and Caregivers / Parents et Professionnels

Moderator / Modérateur : Emmanuelle Parrenin

Artist musician - Specializing in vocal work

Artiste musicienne - Spécialiste en travail vocal

17.30 - 19.00

ROOM JUSSIEU / SALLE JUSSIEU

WORKSHOP /ATELIER

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

MOVING FORWARD ON INTERNATIONAL COLLABORATIONS

LES AVANCÉES EN MATIÈRE DE COLLABORATIONS INTERNATIONALES

Chair / Modérateur : Walter Kaufmann (Baltimore, USA)

Consensus Building from a Panel of Experts for a Rett Syndrome Severity Scale

A. Bebbington, H. Leonard, J. Downs, A. Anderson (Perth, Australia)

Rett Syndrome: From Patient to Parents- Through Parents to Knowledge. Polish – Belgian Experiences from Medical Geneticist's Point of View

A. Midro, E. Smeets (Bialystok, Poland - Maastricht, the Netherlands - Leuven, Belgium)

Collaboration in Science and Support for Rett Syndrome

R. Overton, A. Clarke, Y. Milne (Luton, Cardiff, UK)

Panel Discussion

17.30 - 19.00

ROOM ODEON / SALLE ODEON

WORKSHOP / ATELIER

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

OSTÉOPOROSIS

OSTÉOPOROSE



A Hand Link from Gene to Care

Aims:

- improve understanding, screening and care of bone demineralization in Rett syndrome

Target Audience: Physicians, caregivers and parents

Moderators: Helen Leonard (Perth, Australia), Aagnès Linglart (Paris, France)

Determinants of Bone Mass in Rett Syndrome

Amanda Jefferson (Perth, Australia)

The Risk of Fracture in Rett Syndrome

Jenny Downs (Perth, Australia)

Bisphosphonates: Paediatric Use and Efficacy in Children with Cerebral Palsy

Véronique Forin (Paris, France)

Conclusion by a Paediatric Surgeon

Philippe Wicart (Paris, France)

With an unrestricted educational Grant from IRIS, Servier France.

17.30 – 19.00

ROOM CONSEIL / SALLE CONSEIL

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

THERAPEUTIC MANAGEMENT

PRISE EN CHARGE THÉRAPEUTIQUE

Chairs / Modérateurs : Ingegerd Witt Engerström (Frösön, Sweden) Meir Lotan (Ariel, Israel)

The Musement: Music/Motor Function

M. Bergström-Isacsson, G. Larsson (Östersund, Sweden)

The Value of Music in Everyday Life Working with Rett Syndrome

M. Bergström-Isacsson (Östersund, Sweden)

Specialised Education from a Multi Professional Prospective

J. Cunningham (Surrey, Uk)

Unveiling Hidden Resources - Communication and Learning in Individuals with Rett Syndrome through Music Therapy

C. Elefant (Bergen Norway)

Rebecca's Story - Positive Change as a Result of Music Therapy for a Four Year Old Child with Rett Syndrome

C. Morison, C. Pullen, J. Cardwell, S. Hackett (Morpeth, Ashington, Alnwick, Uk)

The Snoezelen Approach for Individuals with Rett Syndrome

M. Lotan (Ariel, Israel)

17.30 – 19.00

ROOM POISSY / SALLE POISSY

SATELLITE SYMPOSIUM / SYMPOSIUM SATELLITE

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organized by Edelweiss Association

Organisé par l'Association Edelweiss

THE LEARNING AND THE SCHOOL INCLUSION FOR CHILDREN WITH DISABILITIES: SUCCESS STORIES

L'APPRENTISSAGE ET LA SCOLARISATION DES ENFANTS ET JEUNES POLYHANDICAPÉS : DES EXPÉRIENCES RÉUSSIES

Sunday October 12 – Dimanche 12 Octobre

APPROCHE ÉDUCATIVE EN FRANCE

St Germain en Laye 78 - Témoignage de l'expérience d'apprentissage au sein d'un Institut Médico Educatif- Les chemins de l'éveil accueillant des enfants polyhandicapés
Annie Simonet institutrice Montessorienne

Vaucresson 92- Education spécialisée en complément de l'éducation nationale à travers l'expérience d'une co-présence dans un centre médico-pédagogique et approches des apprentissages cognitifs.

Mary Barhet « (Quelque Chose en plus) et Evelyne Justin-Joseph (INS HEA)

L'atelier des odeurs pour les polyhandicapés.

Marie-Françoise Olsem (INSHEA-92 Suresnes)

Le rôle pivot d'un centre de ressource Multihandicap-

Philippe Rousset (centre ressource polyhandicap Paris)

TÉMOIGNAGE D'UNE MÈRE :

La prise en charge de sa fille dans trois pays européens (Angleterre, France et Allemagne)

Anne Dionisi-Fuong (Edelweiss)

**17.30 – 19.30 ROOM SORBONNE / SALLE SORBONNE
BEST OF THE DAY / « BEST OF » DU JOUR**

LANGUAGE: FRENCH / LANGUE : FRANÇAIS

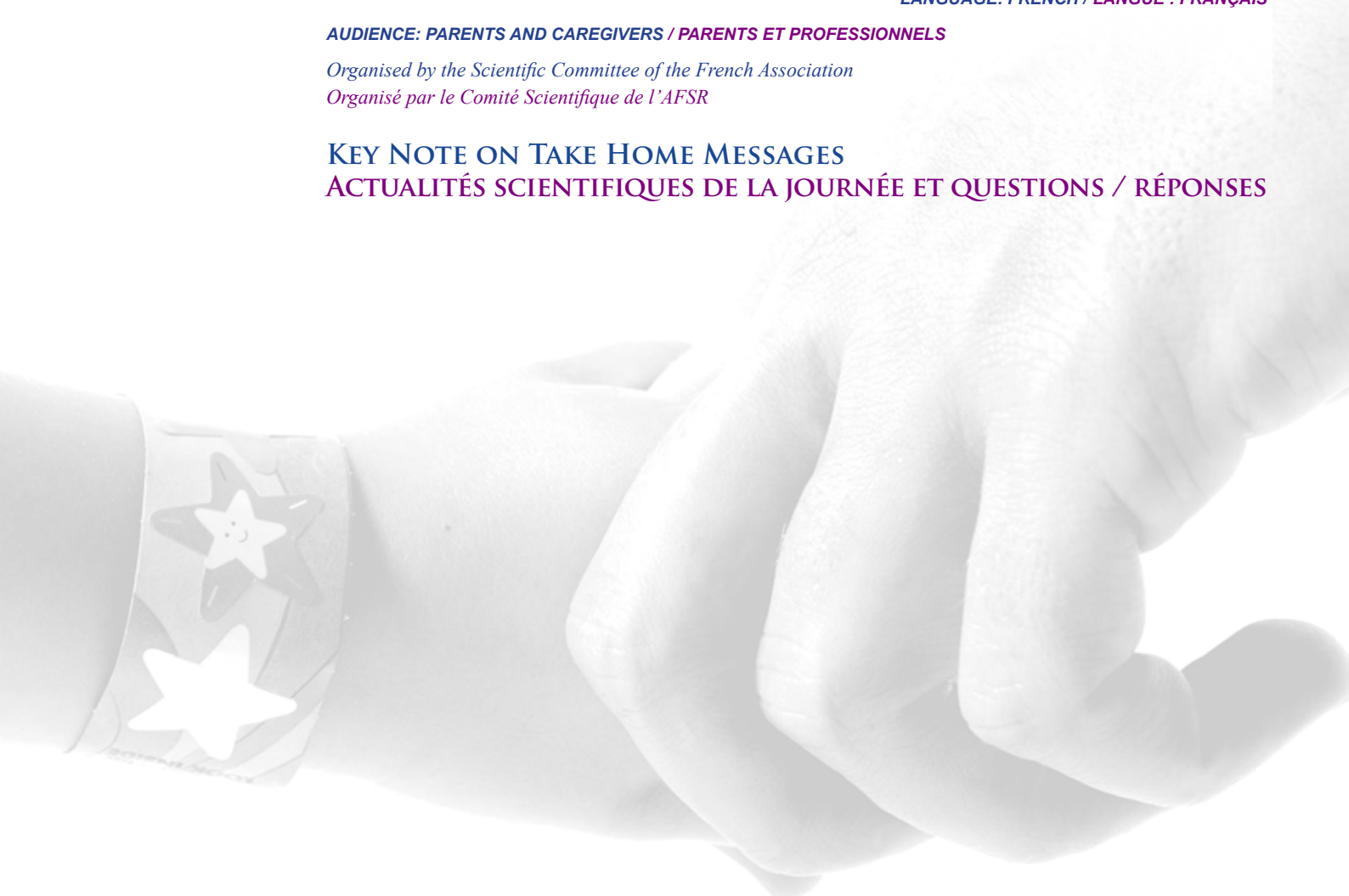
AUDIENCE: PARENTS AND CAREGIVERS / PARENTS ET PROFESSIONNELS

Organised by the Scientific Committee of the French Association

Organisé par le Comité Scientifique de l'AFSR

KEY NOTE ON TAKE HOME MESSAGES

ACTUALITÉS SCIENTIFIQUES DE LA JOURNÉE ET QUESTIONS / RÉPONSES



MONDAY OCTOBER 13 – LUNDI 13 OCTOBRE

8.30 – 10.00

ROOM JUSSIEU / SALLE JUSSIEU WORKSHOP / ATELIER

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

EPILEPSY IN RETT SYNDROME: DIAGNOSIS, CLINICAL AND GENETIC CORRELATES AND MANAGEMENT

EPILEPSIE DANS LE SYNDROME DE RETT : CORRÉLATIONS DIAGNOSTIQUE, CLINIQUE ET GÉNÉTIQUE ET PRISE EN CHARGE

Aims:

- review of specific characteristics of epilepsy in Rett syndrome girls
- correlation with other clinical and genetic data
- treatment approach

Target Audience: Pediatric neurologists, developmental pediatrics, geneticists

Chair: Bruria Ghidoni Ben Zeev (Ramat-Gan, Israel)

Epilepsy in Rett Syndrome in Our Childrens Hospital

Mercedes Pineda (Barcelona, Spain)

Epilepsy in Rett Syndrome

Lakshmi Nagarajan (Perth, Australia)

Epilepsy in Rett syndrome- the Experience of the Israeli Rett Center

Bruria Ghidoni Ben Zeev (Ramat-Gan, Israel)

West Syndrome and Angelman-like Absence Seizures: Atypical Presentations of Epilepsy in Typical RETT syndrome

Nadia Bahi-Buisson (Paris, France)

Therapy of Epilepsy in Rett Syndrome

Peter Huppke (Gottingen, Germany)

8.30 – 10.00

ROOM SAINT GERMAIN / SALLE SAINT GERMAIN ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

CLINICAL MANAGEMENT

PRISE EN CHARGE CLINIQUE

Moderators / Modérateurs: Gordon Baikie (Parkville Vic., Australia), Carolyn Ellaway (Westmead, Australia)

Retrospective Analysis of Patients Attending Australia's First Rett Syndrome Multidisciplinary Management Clinic.

C. Ellaway, Z. Horton, J. Christodoulou, S. Thompson, J. Cowell, R. Kirkland, J. Newsom, B. Bennetts, E. Jay, M. Lopes (Sydney, Australia)

Detection of Alert Factors of Malnutrition Rett Syndrome: A National Prospective Survey within the French National Association (AFSR)

I. Benigni, C. Senez (Laroque-Les-Albères, France)

Factors Affecting Skeletal Integrity in an Australia Rett Syndrome Cohort

A. Jefferson, S. Fyfe, H. Woodhead, J. Briody, A. Bebbington, P. Jacoby, S. Dhaliwal, H. Leonard (Perth, Sydney, Australia)

Increased Fracture Rate and Bone Turn-Over is Associated with Low Bone Mineral Density in Girls with Rett Syndrome

A. Linglart, M. Garabedian, V. Forin (Paris, France)

The Israeli Rett Syndrome Center. Evaluation and Transdisciplinary Play-Based Assessment

M. Lotan, C. Elefant, J. Wine, E. Saraf, Y. Yoshei (Ramat-Gan, Israel)

Rett Syndrome Experience in 62 Mexican Patients Cohort

V. Medina-Crespo, A. Gonzalez-Del Angel, G. Rubio-Rincon (Mexico)

Plasma Leptin Concentrations and Oxidative Stress in Rett Syndrome

J. Hayek, C. De Felice, P. Bardi, A. De Lalla, C. Signorini, S. Leoncini, G. Vonella, L. Ciccoli (Siena, Italy)

8.30 – 10.00

ROOM CONSEIL / SALLE CONSEIL

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

MOLECULAR GENETICS

GÉNÉTIQUE MOLÉCULAIRE

Chairs / Modérateurs: Carolyn Schanen (Wilmington Delaware, USA), Thierry Bienvenu (Paris, France)

MeCP2 Activates the Expression of Nuclear Proteins Associated with Chromatin Remodeling in Vitro

E. Gak, M. Vecsler, A. J. Simon, G. Rechavi, N. Amariglio, E. Schonfeld, M. Segal, B. Ben-Zeev Ghidoni (Tel Hashomer, Tel Aviv, Rehovot, Israel)

Characterization of HIPK2 that, by Associating with MeCP2 Might Function as a Modifier Gene in Rett Syndrome

N. Landsberger, B. Conca, G. Bracaglia, S. Soddu, C. Kilstrup-Nielsen (Busto Arsizio, Rome, Italy)

MeCP2 Contributes to Microtubule Stability through the Tubulin Deacetylation

A. Saxena, R. Scaife, J. Christodoulou, P. Zhang, J. Beaumont, G. Pelka, P. Tam, H. Leonard, M. Kavallaris, D. Ravine (Perth, Sydney, Australia)

Epigenetic Drugs Modulate MeCP2 Dynamics in Living Cells

M. Marchi, M. Becucci, P. Tognini, M. Maffei, N. Landsberger, G.M. Ratto, M. Costa (Pisa, Busto, Arsizio, Italy)

Disruption of the Cellular Regulation of CDKL5 Might be Relevant for Rett Syndrome

C. Kilstrup-Nielsen, L. Giudici, L. Rusconi, I. Bertani, L. Salvatoni, V. Broccoli, N. Landsberger (Busto Arsizio, Milan, Italy)

Proteomic Analysis of CDKL5-Deficient Human Fibroblasts

J. Nectoux, P. Chafey, L. Camoin, C. Broussard, N. Bahi-Buisson, B. Girard, P. Nusbaum, J. Chelly, T. Bienvenu (Paris, France)

8.30 – 10.00

ROOM ODEON / SALLE ODEON

ORAL COMMUNICATIONS / COMMUNICATIONS ORALES

LANGUAGE: ENGLISH / LANGUE : ANGLAIS

MOUSE MODELS 3

MODÈLES DE SOURIS 3

Moderators / Modérateurs: Uta Francke (Stanford, USA), Michael Johnston (Baltimore, USA)

Age and Region-Specific Disturbances of Monoaminergic Systems in the Brain of MeCP2-Null Mice.

M. Santos, T. Summavielle, A. Silva-Fernandes, A. Teixeira-Castro, S. Teixeira, P. Oliveira, N. Sousa, P. Maciel (Minho Braga, Porto, Barcelona, Portugal)



A Hand Link from Gene to Care

Contribution of Fxyd1, a Mecp2 Target Gene, to the Neuropathology of Rett Syndrome
V. Matagne, J. Raber, S. Ojeda (Beaverton, Portland, USA)

Evidence of Central Cholinergic Hypofunction and Rescuing of the Behavioural Phenotype by Postnatal Choline Supplementation in the Truncated MeCP2-308 Mouse Model
L. Ricceri, B. De Filippis, G. Laviola (Rome, Italy)

Tyrosine Hydroxylase Deficits in the Chemoafferent and the Sympathoadrenal Pathway of the Mecp2 Deficient Mice
J.C. Roux, E. Dura, L. Villard (Marseille, France)

Comparative Gene Expression Analysis in the Adrenal Medulla of the MeCP2-Deficient Mouse
E. Dura, L. Villard, J.C. Roux (Marseille, France)

Behavioural Alterations Can be Unveiled in MeCP2-308 Mice Already Soon after Birth
B. De Filippis, L. Ricceri, G. Laviola (Roma, Italy)

10.00 – 10.30

ROOM MONGE / SALLE MONGE
BREAK & VISIT OF THE POSTERS AND THE EXHIBITION
PAUSE, VISITE DES POSTERS ET DE L'EXPOSITION

10.30 – 12.00

ROOM JUSSIEU / SALLE JUSSIEU
PLENARY SESSION / SESSION PLENIERE

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS)

Chairs / Modérateurs : Yoshiko Nomura (Tokyo, Japan), Ingegerd Witt Engerström (Ostersund, Sweden)

Rett Syndrome, View from Emerging Economies
Syndrome de Rett, situation dans les pays émergents
Xi-Ru. Wu (Beijing, China)

Synthesis, Moving from Diagnosis to Therapies
Du diagnostique aux traitements : que retenir ?
Carolyn Schanen (Wilmington Delaware, USA)

12.00 – 12.30

ROOM JUSSIEU / SALLE JUSSIEU
FAMILY ASSOCIATIONS - CONGRESS AWARDS
ASSOCIATIONS DE PARENTS - PRIX DU CONGRÈS

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH AND ITALIAN)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS ET EN ITALIEN)

Moderators / Modérateurs : Philippe Evrard (Paris, France), Helen Leonard (Perth, Australia)

Distinguished Achievement Award / Distinction
Professor Massaya Segawa, Locomotion in Rett Syndrome

Poster Prizes / Prix Poster

12.30 – 13.00

ROOM JUSSIEU / SALLE JUSSIEU
CLOSING CEREMONY / CEREMONIE DE CLOTURE

LANGUAGE: ENGLISH (SIMULTANEOUS TRANSLATION INTO FRENCH)
LANGUE : ANGLAIS (TRADUCTION SIMULTANÉE EN FRANÇAIS)

*Moderators / Modérateurs : Philippe Evrard (Paris, France), Helen Leonard (Perth, Australia),
Christiane Roque (AFSR, France)*

Monday October 13 - Lundi 13 Octobre