RARE NEUROMETABOLIC AND NEUROGENETIC DISEASES IN ADULTS

JOINT FRENCH-ITALIAN WORKSHOP

SIENA, SEPTEMBER 10-11, 2009
RARE NEUROMETABOLIC
AND NEUROGENETIC DISEASES IN ADULTS

JOINT FRENCH–ITALIAN WORKSHOP

French Society of Neurology

Italian Society of Neurology

Presidents

Antonio Federico
Department of Neurological, Behavioural and Neurosurgical Sciences,
Medical School, University of Siena - V.le Bracci 2, 53100 Siena, Italy
federico@unisi.it
tel +39.0577.585763 - fax +39.0577.40327

Olivier Lyon-Caen,
Fédération des Maladies du Système Nerveux, Hôpital de la Salpêtrière,
47 boulevard de l’ Hôpital, 75651 Paris cedex 13 France
olivier.lyon-caen@psl.aphp.fr

Scientific Committee

Giorgio Bernardi, President Italian Society of Neurology
Gioacchino Tedeschi, Secretary of the Italian Society of Neurology
Jean-Marc Léger, General Secretary of the Société Française de Neurologie
Marie-Germaine Bousser, Past President of the Société Française de Neurologie
Jean-Michel Vallat, Past President of the Société Française de Neurologie
Giorgio Cruccu, Coordinator of the Scientific Committee

Local Organizing Committee

Maria Teresa Dotti
Department of Neurological, Behavioural and Neurosurgical Sciences,
Medical School, University of Siena - V.le Bracci 2, 53100 Siena, Italy
dotti@unisi.it
tel +39.0577/585764 - fax +39.0577.40327

Maria Alessandra Carluccio and Daniela Marino
Department of Neurological, Behavioural and Neurosurgical Sciences,
Medical School, University of Siena - V.le Bracci 2, 53100 Siena, Italy
14.00  Opening Ceremony
Welcoming words
Session
A general overview on rare diseases

14.30 European Project for Rare Diseases National Plans Development (EUROPLAN) and the Italian actions on rare diseases
D. Taruscio (Rome)

15.00 The Tuscany network and registry for rare diseases
F. Bianchi, A. Pierini (Pisa)

15.30 The diagnosis of rare diseases in the south of Italy: experience of the CNR Institute of Neurosciences
A. Gambardella (Catanzaro)

16.00 The clinical and laboratory approach to rare neurological diseases
A. Federico (Siena)

16.30 Inborn errors of metabolism in adult neurology: a clinical approach
O. Lyon-Caen, F. Sédel (Paris)

17.00 Coffee break

Update on Mitochondrial encephaloneuromyopathies

17.30 Neuropathies induced by mitochondriopathies
J.M. Vallat (Limoges)

Novel pathogenetic mechanisms in mitochondrial disorders
M. Zeviani (Milan)
Session
Update on Spinocerebellar ataxias (SCAs)

9.00 Dominant ataxias
A. Brice (Paris)

9.30 Autosomal recessive ataxias
A. Filla (Naples)

Update on Small vessel diseases

10.00 Monogenic varieties of small vessel diseases of the brain
M.G. Bousser (Paris)

10.30 CADASIL: Siena experience
M.T. Dotti (Siena)

11.00 Coffee break

Update on late onset Leukodystrophies

11.15 Metachromatic leukodystrophy and Krabbe disease
N. Baumann (Paris)

11.45 Adult-onset non-metabolic leukodystrophies
M.T. Giordana (Turin)

Update on Amyotrophic Lateral Sclerosis

12.15 Update on ALS care
C. Desnuelle (Nice)

12.45 Seeking the causes of ALS: TDP-43 and FUS
V. Silani (Milan)

13.15 Lunch
Session
Update on myopathies

14.30 LGMD with Calpain 3 deficiency: past and present
M. Fardeau (Paris)

14.50 Natural history of calpainopathy and
dysferlinopathy
C. Angelini (Padua)

Update on Charcot-Marie-Tooth disease

15.30 Autosomal recessive Charcot-Marie-Tooth disease
O. Dubourg (Paris)

15.50 Charcot-Marie-Tooth disease: (too) many genes
for several neuropathies
G.M. Fabrizi (Verona)

16.10 CMT1A and 1B: from molecular
pathomechanisms to therapy
A. Schenone (Genova)

Update on dysimmune chronic neuropathies

16.30 CIDP and its variants
E. Nobile-Orazio (Milan)

17.00 Neuropathies associated with monoclonal
gammopathies
J.M. Léger (Paris)
Update on treatable Neurometabolic genetic diseases

17,30 Focus on Glycogenosis
A. Toscano (Messina)
Focus on Fabry’s disease
W. Borsini, S. Büchner (Florence)
Focus on Cerebrotendinous Xanthomatosis
A. Federico (Siena)
Focus on Niemann Pick C disease
F. Sédel (Paris)
Gene therapy in Muscular dystrophy
N. Bresolin (Milan)

18,30 Concluding remarks
A. Federico (Siena), J.M. Léger (Paris)
**Congress Location**
Faculty of Law
Via Mattioli, 10 Siena
Siena, 10 - 11 September 2009

**Language**
English is the official language of the Congress

For inquiries, please contact:

**Local Organizing Secretariat**
Studio ConventurSiena
Via del Cavallerizzo, 1 - 53100 Siena
Tel. 0577 45333 - 0577 270870 - Fax 0577 289334
info@conventursiena.it

**Travel information**
The nearest airports to Siena are Florence or Pisa. Florence is 70 Kms and Pisa 100 Kms. Connections from Florence Airport are by bus to Florence Train Station (15 min) and then there are many busses to Siena every 30-60 min, arriving in the Center of Siena, where are located the Hotels.

From Pisa Airport, there is a frequent train to Florence. You have to change to Empoli for a train to Siena (1h,30). From Siena Train Station to Centers, bus or taxi.

SITA run express buses from Florence to Siena, departing from the SITA bus station (approximately hourly). The trip takes an hour and fifteen minutes. Trains are not such a good option, nearly two hours. If you take the train you will need to catch a local bus from the railway station into the heart of Siena.
Registration Form
Rare neurometabolic and neurogenetic disease in adults Joint French-Italian Workshop Siena, 10-11 September 2009

First name: 
Family name: 
Address: 
ZIP: 
City: 
Country: 
Phone: 
E-mail: 

<table>
<thead>
<tr>
<th>Members of European National Neurological Societies, individual EFNS members</th>
<th>Early Fee (until July 15, 2009)</th>
<th>Late Fee (July 16 / September 1, 2009)</th>
<th>On Site</th>
</tr>
</thead>
<tbody>
<tr>
<td>€ 150</td>
<td>€ 200</td>
<td>€ 250</td>
<td></td>
</tr>
<tr>
<td>Non-members</td>
<td>€ 200</td>
<td>€ 250</td>
<td>€ 300</td>
</tr>
<tr>
<td>Residents and students*</td>
<td>€ 100</td>
<td>€ 100</td>
<td>€ 100</td>
</tr>
</tbody>
</table>

* Student registration form must be accompanied by a letter from the head of department confirming their status and/or a valid student card

Fees for participants and residents include:
- Participation in scientific sessions
- The printed material of the Congress
- A certificate of attendance
- Opening session and welcome reception
- Lunch and coffee during breaks

Payment
Bank transfer to “RIUNIONE ITALO FRANCESE COMITATO ORGANIZZATORE RIUNIONE CONGIUNTA”
Bank details: IBAN IT 16 C 01030 14200 000006574158 - SWIFT PASCITMMSIE
Bank charges are the responsibility of the payer and should be paid in addition

Cancellation policy
- Cancellations received between July 1 and August 9, 2009 - 50% refund will be made
- After August 10, 2009 - no refund will be made

Please return this form by Fax 0039 0577 289334 or by email: info@conventursiena.it with copy of bank transfer

Date _______________ Signature _____________________
Accommodation Form
Rare neurometabolic and neurogenetic disease in adults Joint French-Italian Workshop Siena, 10-11 September 2009

Please book:

- [ ] +++ Hotel
- [ ] ++++ Hotel
- [ ] Bed and Breakfast
- [ ] Single Room
- [ ] Double Single Occupancy
- [ ] Double Room

Date of arrival ___________________ Date of departure ___________________
Accompanying person: __________________________________________

<table>
<thead>
<tr>
<th>HOTEL DEPOSIT</th>
<th>€ 60</th>
</tr>
</thead>
<tbody>
<tr>
<td>RESERVATION FEE</td>
<td>€ 10</td>
</tr>
<tr>
<td>TOTAL</td>
<td>€ 70</td>
</tr>
</tbody>
</table>

Payment
Bank transfer to “RIUNIONE ITALO FRANCESCO COMITATO ORGANIZZATORE RIUNIONE CONGIUNTA”
Bank details: IBAN IT 16 C 01030 14200 000006574158 - SWIFT PASCITMMSIE
Bank charges are the responsibility of the payer and should be paid in addition

Please return this form by Fax 0039 0577 289334 or by email: info@conventursiena.it with copy of bank transfer

I authorize the processing of my personal data in compliance with the Italian Data protection Law in force (D.Lgs. 196/2003)

Date ______________ Signature _______________________________
We thank:

Actelion Pharmaceuticals Italia
Ebewe Pharma
Genzyme
Kedrion
Sigma Tau

Bancasciano
Banca di Monteriggioni Credito Cooperativo
Cover illustration: Yves Brayer (France, 1907-1990)