



**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**

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### **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

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Maria Alessandra Carluccio and Daniela Marino

Department of Neurological, Behavioural and Neurosurgical Sciences,  
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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:		

	Early Fee (until July 15, 2009)	Late Fee (July 16 / September 1, 2009)	On Site
Members of European National Neurological Societies, individual EFNS members	€ 150	€ 200	€ 250
Non-members	€ 200	€ 250	€ 300
Residents and students*	€ 100	€ 100	€ 100
* 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](mailto: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: .....

HOTEL DEPOSIT	€ 60
RESERVATION FEE	€ 10
<b>TOTAL</b>	<b>€ 70</b>

### Payment

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*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

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