

# 9<sup>TH</sup> International Symposium on Mucopolysaccharide and Related Diseases

9<sup>o</sup> Simposio Internazionale sulle Mucopolisaccaridosi e Malattie Affini  
29 giugno - 2 luglio, 2006



**SCIENTIFIC AND FAMILY PROGRAMMES**

## Scientific Committee

M. Scarpa, Padua, Italy (Chair)  
G. Andria, Naples, Italy  
A. Ballabio, Naples, Italy  
M. Beck, Mainz, Germany  
G. Coppa, Ancona, Italy  
R. Desnick, New York, USA  
P. Di Natale, Naples, Italy  
M. Di Rocco, Genoa, Italy  
M. Filocamo, Genoa, Italy  
J.J. Hopwood, Adelaide, Australia  
I. Maire, Lyon, France  
J. Muenzer, Chapel Hill, USA  
E.F. Neufeld, Los Angeles, USA  
R. Parini, Monza, Italy  
E. Paschke, Graz, Austria  
C. Peters, Kansas City, USA  
R. Ricci, Rome, Italy  
A. Rovelli, Monza, Italy  
W.S. Sly, St. Louis, USA  
R. Tomanin, Padua, Italy  
S. Tomatsu, St. Louis, USA  
B. Winchester, London, UK  
E. Wraith, Manchester, UK  
E. Zammarchi, Florence, Italy  
F. Zacchello, Padua, Italy

scientific.committee@congress2006mps.it

## Organizing Committee

Italian MPS Society (Charity)  
Via Savona 13  
20144 Milano

Department of Pediatrics, University of Padua  
Via Giustiniani, 3  
35128 Padova

organising.committee@congress2006mps.it

## Congress Organization

Salus Pueri Biomedicine srl  
Via Marconi 30  
50131 Firenze

congress.organisation@congress2006mps.it

Daniela Roselli  
Elena Ferri  
Chiara Zanetti

## Web Site

The programme is available on the web site [www.congress2006mps.it](http://www.congress2006mps.it)

## Credits:

- **ECM: 13 credits for Medical Personnel , 12 credits for Nurse Personnel**  
- **EACCME/UMS: 18 credits**



**14.30-15.00 OPENING ADDRESSES AND WELCOMES**

**15.00-17.00 CLINICAL ASPECTS**

*Chair: M. Beck, Germany; G.V. Coppa, Italy*

- 15.00-15.20 Epidemiology of the mucopolysaccharidoses and related disorders (J.J. Hopwood, Australia)  
15.20-15.40 Clinical variability. Is prediction of clinical severity possible in the single case? (J. Muenzer, USA)  
15.40-16.00 Follow up of patients and limitations of current therapies (E. Wraith, UK)  
16.00-16.20 The International MPS Network (C. Lavery, UK)

Discussion

**17.00-17.45 PLENARY LECTURE**

**PROGRESS AND CHALLENGES IN UNDERSTANDING AND TREATING THE MUCOPOLYSACCHARIDOSES (E.F. Neufeld, USA)**

**18.00-19.00 Meeting other Families**

**19.00-20.00 Get together party**

**8.30-10.30 PATHOGENESIS OF LYSOSOMAL STORAGE DISORDERS**

*Chair: B. Winchester, UK; P. Di Natale, Italy*

- 8.30-8.50 Pathophysiology of lysosomal storage disorders: cascade events to irreversible pathology (S.U. Walkley, USA)  
8.50-9.10 Microarrays to study pathologic mechanisms of lysosomal storage disorders (E.H. Schuchman, USA)  
9.10-9.30 Identification and use of molecular markers for lysosomal storage disorders (T. Cox, UK)  
9.30-9.50 Single gene disorders are complex disorders (K. Dipple, USA)

**SELECTED ORAL PRESENTATIONS**

- 9.50-10.00 Identification of serum heparin cofactor II-thrombin complex as a disease biomarker in MPSI (L. Clarke, Canada)  
10.00-10.10 A novel lysosomal disease with granular storage material is caused by cathepsin D deficiency (R. Steinfeld, Germany)  
10.10-10.20 Heparan sulfates released in MPSIIIB patient urines activate inflammatory response through TLR4 receptor (J. Ausseil, France)  
10.20-10.30 Molecular pathology of mucopolysaccharidosis IIIB (G.R.D. Villani, Italy)  
10.30-11.00 Coffee break

**11.00-13.00 NEONATAL SCREENING AND DIAGNOSIS OF LYSOSOMAL STORAGE DISORDERS**

*Chair: J.J. Hopwood, Australia; M. Scarpa, Italy*

- 11.00-11.10 New technologies for the diagnosis of lysosomal storage disorders (J.J. Hopwood, Australia)  
11.10-11.30 Multiplex analysis of dried blood spots using tandem mass spectrometry for newborn screening of lysosomal storage disorders (M. Gelb, USA)  
11.30-11.50 Multiplex protein profiling (P.J. Meikle, Australia)  
11.50-12.10 New tools for the diagnosis of lysosomal storage disorders (M. Fuller, Australia)  
12.10-12.30 Screening of lysosomal storage disorders: pilot study (G. Nijzawa, USA)  
12.30-12.50 Screening and ethical problems (F. Stewart, Ireland)

**SELECTED ORAL PRESENTATIONS**

- 12.50-13.00 Discovery of biomarkers for mucopolysaccharidosis types I, IIIA and IIIB by SELDI-TOF mass spectrometry and their use in monitoring enzyme replacement therapy in MPS I patients (C. Beesley, UK)

**9.00-10.45 EMERGENCY AND HEART PROBLEMS***Discussants: M.Beck, Germany ; M. Di Rocco Italy*

Best practice in anesthesiology (R. Walker, UK)

A family facing anesthesia (F. Bertoglio, Italy)

Understanding the heart: cardiac complications in mucopolysaccharide storage diseases at long-term follow-up (V. Fesslova, Italy)

I had a valve replacement but..... (A. Serafini, Italy)

Bleeding and clotting problems (A. Vellodi, UK)

**10.45-11.00** Coffee break**11.00-12.30 RESPIRATORY PROBLEMS***Discussants: E. Wraith, UK; G. Parenti, Italy*

The respiratory tract in patients with mucopolysaccharidosis type I (MPSI) (W. Kamin, Germany)

My child has a tracheostomy (R. Di Lollo, Italy)

My son is using a CPAP (B. Wildi, Germany)

Managing ENT problems (A. Keilmann, Germany)

**PROFESSIONALS****13.00-14.30 GENZYME Sponsored Workshop****CONCILIATE SCIENTIFIC EVIDENCE AND INDIVIDUAL EXPERIENCE WITH ALDURAZYME®****14.30-16.00 ANIMAL MODELS***Chair: S. Tomatsu, USA; A. Ballabio, Italy*

14.30-14.50 Animal models for lysosomal storage disorders (M. Haskins, USA)

14.50-15.10 A canine model for MPSIIIB (N.M. Ellinwood, USA)

15.10-15.30 Use of animal models for unraveling mechanisms of pathogenesis in lysosomal storage disorders (A. D'Azzo, USA)

**SELECTED ORAL PRESENTATIONS:**15.30-15.40 Murine model (*Galns<sup>tm(C76S)slu</sup>*) of MPS IVA with a missense mutation at an active site among sulfatase proteins (S. Tomatsu, USA)

15.40-15.50 Life without sulfatases: exploiting a mouse model of multiple sulfatase deficiency (C. Settembre, Italy)

15.50-16.00 Morphopathological features in tissues of  $\alpha$ -mannosidosis guinea pigs at different gestational ages. (D. Auclair, Australia)**16.00-16.30** Coffee break

Poster viewing

**16.30-18.00 ACTELION Sponsored Workshop****LONG-TERM MANAGEMENT OF PATIENTS WITH GAUCHER DISEASE TYPE I: PROGRESSING BEYOND CURRENT PARADIGMS****FAMILIES****15.00-16.15 FEEDING PROBLEMS***Discussants: N. Guffon, France; R. Parini,*

About gum and teeth problems in mucopolysaccharidoses (P. Mariuzza, Italy)

Assessment of feeding difficulties in children with Sanfilippo disease (MPS III) (M. Ryan, UK)

Why have a gastrostomy? (M. Di Rocco, Italy)

My daughter has a gastrostomy.... (F. Pidden, UK)

**16.15-16.30** Coffee break

**16.30-17.45 ORTHOPEDIC AND SKELETAL PROBLEMS**

*Discussants :S. Tomatsu, USA; M. De Pellegrin, Italy*

Spine, hips and knees: Indication experience and results (C. Gigante, Italy)

Bracing of the spine and surgery of the big joints in Mucopolysaccharidosis (B. Link, Germany)

Should my child have physiotherapy for joint stiffness? (L. Tedesco, Italy)

Quality of life with Morquio (G. Caspani, Italy)

I have Morquio and I am at University (J. Evans, UK)

International Morquio A Registry (S. Tomatsu, USA)

**19.30 Classical concert and “Life for MPS Award”**

**Saturday July 1, 2006**

**FAMILIES AND PROFESSIONALS****8.30-10.30 THERAPY FOR MUCOPOLYSACCHARIDOSES: Update and Perspectives**

*Chair: W.S. Sly, USA; G. Andria, Italy*

8.30-8.50 Summary and update of clinical trials for MPSI (E. Wraith, UK)

8.50-9.10 Summary and update of clinical trials for MPSII (J. Muenzer, USA)

9.10-9.30 Long-term enzyme replacement therapy with Naglazyme for MPSVI (Maroteaux-Lamy syndrome): growth, pubertal development, and pulmonary function (P. Harmatz, USA)

9.30-9.50 Therapy for Sanfilippo syndrome (K.M. Hemsley, Australia)

9.50-10.05 Perspectives for the therapy of MPSIV (S. Tomatsu, USA)

10.05-10.20 Perspectives for the therapy of MPSVII (W.S. Sly, USA)

10.20-10.40 Studies to enable the monitoring of efficacy resulting from therapies (M. Beck, Germany)

**10.40-11.00** Coffee break

**11.00-13.00 HEMATOPOIETIC STEM CELL TRANSPLANTATION AND ALTERNATIVE THERAPIES**

*Chair: C. Peters, USA; A. Rovelli, Italy*

11.00-11.20 Outcome of haematopoietic cell transplantation for Hurler's Syndrome: European experience 1994-2004 (J.J. Boelens, The Netherlands)

11.20-11.40 Therapeutic potential of stem cells (P. Gupta, USA)

11.40-12.00 Psychosocial outcomes of BMT for MPSIH: patient self-report of personality and personal adjustment (N. Wager, UK)

12.00-12.20 Substrate reduction therapy for glycosphingolipid lysosomal storage disorders (F. Platt, UK)

12.20-12.40 Pharmacologic chaperone therapy for lysosomal diseases (R. Desnick, USA)

**SELECTED ORAL PRESENTATIONS:**

12.40-12.50 Gene expression-targeted isoflavone therapy for Sanfilippo disease: in vitro studies on fibroblasts and a pilot clinical trial (G. Wegrzyn, Poland)

12.50-13.00 Is stop codon read-through a viable strategy for treating lysosomal storage disorder patients? (D.A. Brooks, Australia)

**PROFESSIONALS****13.00-14.30 SHIRE HUMAN GENETIC THERAPIES Sponsored Workshop  
UPDATE ON HUNTER SYNDROME (MPS II)****14.30-16.30 WORKSHOP ON THE BLOOD-BRAIN BARRIER**

*Chair: J. Muenzer, USA; M. Scarpa, Italy*

14.30-14.40 The Blood-Brain Barrier in health and disease (H. Wolburg, Germany)

14.40-15.10 Delivery of therapeutic agents to the CNS: the problems and the possibilities and the relevance for lysosomal storage disorders (D.J. Begley, UK)

15.10-15.30 Blood brain barrier in neuro-degenerative diseases (A. Leon, Italy)

**SELECTED ORAL PRESENTATIONS:**

15.30-15.40 CNS delivery of rhNS in the MPSIIIA Huntaway dog following cisternal injection (A.C.Crawley, Australia)

15.40-15.50 Intrathecal enzyme replacement therapy for MPSI: beyond the laboratory (P. Dickson, USA)

15.50-16.00 CSF GAG behavior during intrathecal enzyme replacement therapy in a patient with MPS I and spinal cord compression (M.V.R. Munoz, USA)

16.00-16.10 Histopathological and behavioral improvement of murine mucopolysaccharidosis Type VII by intracerebral transplantation of neural stem cells (T. Okuyama, Japan)  
Poster viewing

**16.30-18.00 BIOMARIN Sponsored Workshop  
NEW FRONTIERS IN MPS**

**18.00-19.30 GOLD, GLOBAL ORGANISATION FOR LYSOSOMAL DISEASES  
ANNUAL GENERAL MEETING.**

Guest Lecture "Medicine for lysosomal diseases: Past imperfect, future tense" (T. Cox, UK)

**FAMILIES**

**15.00-16.30 EYE PROBLEMS AND NEUROLOGICAL ASPECTS**

*Chair Discussants: R. Giugliani, Brazil, E. Zammarchi, Italy*

The dilemma of eyes (S. Pitz, Germany)

Achieving my full educational potential (M. Broughton, UK)

Recognizing signs and symptoms of neurological problems (E.P. Sganzerla, Italy)

How should I treat my child with epilepsy? (D. Grioni, Italy)

My child does not sleep....What can I do?... (F. Bertoglio, Italy)

Behavioural phenotype in MPS and related disorders – Implications for research and intervention (P. Santosh, UK)

**16.30-16.45** Coffee break

**16.45-18.00 PSYCHOLOGICAL ASPECTS**

*Discussants: L. Clarke, Canada; L. Pavone, Italy*

MPSI: the disease from the point of view of the patient (J.L. Diaz, USA)

Pain incidence in children with lysosomal storage disorders (F. Reiter, France), (C. Agosto, Italy)

Living with MPS and waiting for a therapy (B. Baldi, Italy)

Living with Sanfilippo disease (P. Hawkins, UK)

My brother and sister have Sanfilippo disease (H. Donegani, UK)

I passed on Hunter Disease..... (G. Minacco, Italy)

Facing the loss of our children (E. Volk, Germany)

**Summer Dinner**

**Sunday July 2, 2006  
PROFESSIONALS**

**8.30-10.30 GENE THERAPY AND LATE BREAKING NEWS**

*Chair: E.F. Neufeld, USA; R. Tomanin, Italy*

8.30-8.50 Gene therapy of the brain in the dog model of Hurler syndrome (J.M. Heard, France)

8.50-9.10 Gene therapy for Mucopolysaccharidosis (K. Ponder, USA)

**SELECTED ORAL PRESENTATIONS**

9.10-9.20 Correction of established neurologic disease and evidences of in vivo cross correction in the mouse model of metachromatic leukodystrophy (A. Biffi, Italy)

9.20-9.30 AAV-mediated gene transfer to muscle and liver of MPSVI animal models (A. Tessitore, Italy)

9.30-9.40 Therapeutic impacts of combining an intravenous and an intracisternal delivery of AAV2 vector on MPS IIIB in MICE Haiyan (A. Fu, USA)

9.40-9.50 AAV-mediated delivery of sulfamidase and SUMF1 genes to the brain of MPSIIIA mice: a therapeutic strategy for MPSIIIA (A. Fraldi, Italy)

9.50-10.00 Inhibition of the EXT1 gene expressions by RNA interference as a therapeutic approach for Sanfilippo A syndrome, (D. Grinberg, Spain)

10.00-10.10 Therapeutic effect induced in MPSI mouse model by intraperitoneal implantation of alginate microcapsule over-expressing alpha-l-iduronidase (IDUA) (E. Piller Puicher, Italy)

**10.10.-10.30** Late breaking news

## **FAMILIES**

### **8.30-10.30      LOOKING TO THE FUTURE**

*Chair: C. Lavery, UK; F. Bertoglio, Italy*

Communication from the International MPS Network

Experiences from MPS Associations: Round Table (Kirsten Harkins, Canada; B. Wedehase, USA, and MPS delegates)

What should we do to increase knowledge on MPS? (G. Pastores, USA)

## **FAMILIES AND PROFESSIONALS**

### **11.00-12.00      SUMMARY OF THE SYMPOSIUM**

**G. Andria, Italy**

**Closing Remarks**

**.....PROPOSALS FOR THE 2008 SYMPOSIUM**



## ***THE FANCY SIDE OF THE PROGRAMME***

### VENETIAN WINE TASTING

SPONSORED BY "SANTA MARGHERITA WINE GROUP", Fossalta di Portogruaro, in collaboration with the Italian Sommelier Association-Veneto (AIS-Veneto)

Each tasting session will host 30-40 participants.  
Booking will be accepted at the Conference Center.

FRIDAY 30<sup>TH</sup> 2006

11.00-12.00

17.00-18.00

WHITE AND SPARKLING WINES



SATURDAY JULY 1ST 2006

11.00-12.00

17.00-18.00

ROSE', RED AND PASSITOS WINES

"SANTA MARGHERITA WINE GROUP"

Santa Margherita is part of a group of widely-diversified businesses, based in the Eastern Veneto region and controlled by the Marzotto family.

Santa Margherita has over 50 year experience in producing ultramodern wines, with a complete range of still and sparkling wines made from grapes traditionally grown in the Trentino-Alto Adige and Eastern Veneto regions.

The company owes its success to its unfailing ability to predict consumers' changing tastes, and to supply the market with top-quality, yet easy-to-drink wines. The whites are fresh-tasting with a fragrant aroma while the reds are full-bodied and soft.





## ***THE CULTURE SIDE OF THE PROGRAMME***

***CLASSIC CONCERT S. GIORGIO CHURCH, VENICE***

***FRIDAY JUNE 30TH 2006***

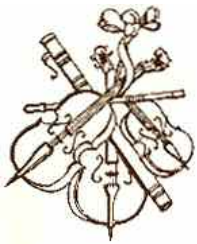
7.30 pm

Ensemble: I Solisti Veneti

Conductor: Claudio Scimone

Music by: JS. Bach, A. Vivaldi , G. Rossini, N. Paganini

During the Concert The "Life For MPS Award" will be presented.



"I Solisti Veneti" directed by Claudio Scimone enjoy a firm reputation both with the general public and with the specialist music press as the most popular and technically outstanding Italian orchestra and one of the finest orchestras in the world, a reputation based not only on their highly acclaimed concert performances and recordings but also on the impressive variety of the cultural and artistic fields they have explored.

Founded by Claudio Scimone in Padua in 1959, "I Solisti Veneti" have given concerts in over sixty countries in Europe, North and South America, Japan, Australia and elsewhere. They make regular appearances at the world's leading Festivals, including, since 1965, the Salzburg Festival (every year from 1965 until 1998). "I Solisti Veneti" have recorded more than 300 LP and CD for the greatest multinational Companies (Erato-WEA, BMG-RCA, CBS, Frequenz).