



International Association of Medical Genetics (MAGI)  
38066 Riva del Garda (TN), Via Pola N°6

**MAGI ASSOCIATION SECOND INTERNATIONAL CONGRESS**  
**SCIENTIFIC PROGRESS IN DIAGNOSIS AND THERAPY OF GENETIC AND RARE DISEASES**

19th and 20th October 2007

Trento (Italy)

**Introduction:**

On 19-20th October, the International Association of Medical Genetics MAGI will hold its second international congress for researchers, doctors and associations concerned with genetic and rare diseases. In line with the European vocation of the founders of MAGI, the congress will bring together experts from all of Europe, including eastern European countries. For the latter, funds have been earmarked to enable a limited number of researchers to participate free of charge with reimbursement of travel and accommodation expenses.

Most genetic diseases are rare and severely invalidating. Because their onset occurs at birth or in childhood, medical intervention must be organized in a transnational network that enables samples for diagnostic tests, therapeutic protocols and knowledge for well orientated top level research to be transferred rapidly.

This year the accent of the congress will be on diagnostic discoveries and advances in therapeutic protocols (new surgical techniques, use of stem cells, gene therapy and rehabilitation) and on the work of research centres and drug companies for the production of orphan drugs. A highlight will be the opening lecture by Prof. Lucio Luzzatto on new therapeutic approaches to cancer.

European representatives of the Orphanet consortium and of the agency for the development of European projects APRE will be present at the congress to illustrate the European network of rare diseases and applications for 7th Programme European research grants.



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## DAY 1

### First Session

#### 9.00-10.00 Introduction

- Introduction by the founding members of MAGI Association  
Introduzione dei membri fondatori della associazione MAGI
- Presentation of Trentine Rotary Club project for MAGI Association  
Presentazione del Progetto Rotary Club del Trentino per la Associazione MAGI
- Welcome by local authorities  
Saluto della Autorità locali
- Lecture: Management of health service in the molecular medicine era  
Lettura: Gestione del servizio sanitario nell'era della medicina molecolare  
*Dr. Mario Magnani Consiglio Regionale del Trentino Alto Adige*

### Scientific Section

#### 10.00-12.00 Oncology

- Lettura magistrale: Approcci terapeutici al cancro  
Opening lecture: Therapeutic approaches to cancer  
*Prof. Lucio Luzzatto, Direttore Scientifico dell'Istituto Toscano Tumori*
- Studio epidemiologico e genetico di una famiglia allargata protetta dal cancro  
Epidemiology and genetics of an extended family with cancer protection  
*Dottor Roberto Luongo, Associazione Riva del Garda (TN)*
- PARP-1: quale ruolo nella salute e nella malattia?  
PARP-1: Role in health and disease?  
*Professoressa Silvia Sestini, Dpt Biologia Molecolare, Università di Siena*
- Studio clinico e prevenzione nelle famiglie con sindrome di Li-Fraumeni  
Clinical study and prevention in families with Li-Fraumeni syndrome  
*Dottor Alberto Bottini, Ospedale di Cremona*

#### 12.00-13.00 Cystic fibrosis

- Esperienza del centro trentino per la cura della fibrosi cistica  
Experience of the Trentino Cystic Fibrosis Centre  
*Dottor Ermanno Baldo, Ospedale Civile Rovereto*  
Genotipizzazione della pseudomonas nei pazienti affetti da fibrosi cistica
- Genotyping of Pseudomonas in cystic fibrosis patients  
*Dottoressa Alessandra Bragonzi European Institute for Cystic Fibrosis Research Institute H.S. Raffaele Milan*
- Contributo della genomica comparata e funzionale alla comprensione della patogenesi microbica  
Contribution of comparative and functional genomics to bacterial pathogenesis  
*Professor Olivier Jousson, Faculty of Sciences, University of Trento*

#### 13.30-14.30 Lunch



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## **Second Session**

### **14.30-16.30 Neurology**

- Cellule staminali autologhe: applicazioni cliniche nella Sclerosi Laterale Amiotrofica  
Autologous stem cells: clinical applications in Amyotrophic Lateral Sclerosis  
*Prof.ssa Letizia MAzzini, Department of Neurology "Eastern Piedmont University" Novara*
- Diagnostic approach to unexplained hypouricemia  
*Dr. Ivan Sebesta, Inst. Inher. Metab. Dis., First Faculty of Medicine, Charles University Prague, Czech Republic*
- Microarray gene expression profiling of inherited metabolic diseases  
*Prof. Tony Marinaki, Purine Research Laboratory, Guy's Hospitals London, UK*
- Cause dell'Ictus Giovanile  
Causes of juvenile stroke  
*Dr Gian Paolo Anzola, Ospedale Sant'Orsola Brescia*
- Controllo dell'RNA nei neuroni: proteine che legano l'RNA e patologie  
Controlling neuronal RNA: RNA-binding proteins and disease  
*Prof. Paolo Macchi, Center for Brain Research, University of Vienna*

### **16.30-17.30 Cardiology**

- Il ruolo della genetica molecolare nella pratica cardiologica quotidiana  
The role of molecular genetics in everyday cardiological practice  
*Dr Marini Massimiliano APSS Trento*
- Stato dell'arte della cardiologia interventistica nella patologia dei difetti del setto interatriale  
State of the art of Interventional Cardiology in Atrial Septal Defects  
*Dr Eustaquio Onorato Ospedale Sant'Orsola Brescia*

### **17.30-18.00 European projects**

- Il 7° programma quadro: Tema salute  
7th Framework: Health  
*Dott.ssa Diassina Dimaggio, Agenzia APRE, Roma*

## **DAY 2**

### **First session**



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#### **9.00-11.30 Clinical genetics**

- An European database for rare diseases: the experience of Orphanet  
*Dott.ssa Ana Rath Orphanet Consor Paris*
- Presentazione del servizio di informazione sulle malattie rare dell'Istituto Mario Negri  
The Rare Diseases Information Center of the Mario Negri Institute  
*Dottor Luca Barcella, Center for rare diseases "Aldo e Cele Dacco, Mario Negri Institute*
- Consulenza teratologica sugli effetti dei farmaci assunti in gravidanza  
Teratological counselling on effects of drugs taken during pregnancy  
*Dott.ssa Silvia Belli, Servizio di consulenza genetica APSS Trento*
- Terapia odontoiatrica nei pazienti con malattie genetiche e rare  
Dental therapy in patients with rare genetic diseases  
*Dottor Valesi Penso, APSS Trento*
- The gene therapy approach for inherited skin blistering diseases  
*Prof. Guerrino Meneguzzi Faculté de Médecine Université de Nice, France*

#### **11.30-13.30 Medical biotechnologies**

- Alterazioni dell'mRNA editing riscontrate in malattie neurologiche  
Changes in mRNA editing in neurological diseases  
*Dottor Matteo Bertelli, Associazione Riva del Garda (TN)*
- Translational profiling: a new systems-based perspective  
"Profili traduzionali: una nuova prospettiva di biologia dei sistemi"  
*Professor Alessandro Quattrone, Università di Trento*
- Studio del proteoma nelle patologie umane  
Proteomics in human diseases  
*Professoressa Annalisa Santucci Dpt Biologia Molecolare, Università di Siena*
- HLA e Malattie  
HLA and diseases  
*Dottor.ssa Nadia Ceschin e Prof. Paolo Gottardi, APSS Trento*
- Le sindromi paraneoplastiche neurologiche  
Neurological paraneoplastic syndromes  
*Dottor Lorenzo Lo Russo, Ospedale Mellino Mellini di Chiari (BS)*
- Spinal cord motor neuron differentiation, connectivity, and regeneration  
*Prof. Stefano Stifani, McGill University Montreal Neurological Institute, Quebec Canada*

#### **13.30-14.30 Lunch**

### **Second Session**

#### **14.30-15.45 Preventive medicine**



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- Suitable nutrition as an important tool against civilization diseases: soy products  
*Prof. Ing. Jan Káš, Dr.Sc., Dept. of Biochemistry and Microbiology, ICT Prague*
- La dieta in soggetti con malattie metaboliche ereditarie: l'esempio delle iperuricemie
- Diet for hereditary metabolic disease patients: the example of hyperuricemia  
*Professor Giuseppe Pompucci, Università di Siena*

***16.00-17.00 Biotechnological research and development***

- Sviluppo delle aziende biotecnologiche in Trentino
- Development of biotech firms in Trentino  
Dott.ssa Renata Diazzi director of CEII Trento
- L'esperienza della MAGI'S LAB S.r.l.  
The experience of MAGI'S LAB S.r.l.  
Dottor Matteo Bertelli, Associazione MAGI onlus & MAGI'S LAB S.r.l.

***17.00-18.00 Prize giving and conclusion***

**SOCIAL ACTIVITIES**

**Friday 19th October (separate registration)**

21.00-22.30 Gala Dinner for speakers and guests