



“The role of the Catholic clinical network in combatting rare genetic diseases in Europe”

13th June 2013

Headquarters of the European Region Tyrol - South Tyrol - Trentino
45-47 Rue de Pascale, Brussels

The meeting brings together doctors and delegations from centres for research, diagnosis and the pharmaceutical industry with those of the Italian Ministry for Research and Health, European Patients' Organisations, European research, industry and healthcare and the European Consortium for Rare Diseases, Orphanet. The topic is creation of a network of expertise to extend and improve scientific knowledge and healthcare for rare genetic diseases in Europe. For many years, MAGI has been investing resources to strengthen network relationships and build understanding of rare diseases in the European supranational context.

The congress begins with the topic of diagnosis of rare genetic diseases in Italy, highlighting MAGI's role in creating a network of diagnostic and healthcare institutions of Catholic inspiration. MAGI collaborates with the Ospedale Casa Sollievo della Sofferenza at San Giovanni Rotondo, the Policlinico Universitario Agostino Gemelli in Rome and the Ospedale San Giovanni Battista in Rome. For certain genetic diseases, such as Mendelian-inherited lymphedema, MAGI is the only organisation in Italy offering diagnosis; for others, such as hereditary retinal dystrophies, it is one of very few.

A second theme is research in the field of rare genetic diseases. An example of research into Mendelian-inherited stroke and vascular malformations will be presented. Both studies were conducted by MAGI and are the biggest and most complete conducted in Italy.

Closing topics regard the European network for the diagnosis of rare diseases, the diagnostic situation in the Euroregion and the question of prenatal diagnosis to identify fetuses eligible for high-tech therapeutic protocols based on stem cells or gene therapy.

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PROGRAMME:

9am-12.30pm

- **Welcoming addresses:** Representatives of the Euroregion, President of Trento Province, President of Bolzano Province, Captain of Tyrol; Apostolic Nuncio of Belgium, Mons. Giacinto Berloco, Europarliamentarians
- **Dr. Matteo Bertelli**, President of MAGI:
“In search of genetic diseases not yet diagnosed - hope for sufferers of rare diseases”
- **Discussion:** *“The role of patients associations and specialist doctors in building a European network of rare genetic diseases: experience and prospects”*

Presided by:

- **Prof. Leduc Albert**, Dist. Hon. Pres., European Society of Lymphology
 - **Prof. Pierre Bourgeois**, Pres. Scientific Commission, European Society of Lymphology
 - **Prof. Francesco Boccardo**, Vice-Pres., European Society of Lymphology
 - **Dr. Simona Bellagambi**, UNIAMO delegate of EURORDIS Council of Alliances
 - **Dr. Ana Rath**, Editorial Team Manager ORPHANET
 - **Representative of European Research**
 - **Representative of Health and Consumers**
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- **Albrecht Freiherr von Boeselager**, Grand Hospitaller:
“The Order of Malta in Europe: from patient care to application of the evangelical mandate!”
 - **Prof. Sandro Michelini**, Director, San Giovanni Battista Hospital of the Order of Malta:
“Title to be decided: venous malformations and primary lymphedema”



12.30-1.30 pm

- Buffet Lunch

1.30-4pm

- **Dr. Domenico Crupi**, Director General, Casa Sollievo della Sofferenza:
“Casa Sollievo della Sofferenza - a Catholic hospital dedicated to genetic diseases”
- **Dr. Maurizio Scarpa**, Coordinator Rare Diseases – IRCCS “Casa Sollievo della Sofferenza”, San Giovanni Rotondo:
“A clinical service for rare diseases in southern Italy”
- **Prof. Alessandro Pezzini**, Dept. Clinical and Experimental Science, Neurology Clinic, University of Brescia:
“Juvenile stroke and the role of Mendelian-inherited syndromes”
- **Prof. Raul Mattassi**, Angiology and Vascular Surgery Dept., Clinica Humanitas Mater Domini:
“Clinical and genetic study of congenital vascular malformations”
- **Dr. Patrick Willems**, Gendia Network:
“State of the art of the European network for diagnosis of genetic diseases”
- **Prof. Giuseppe Noia**, Policlinico Gemelli:
“The future of prenatal diagnosis: early identification of fetuses to treat with gene therapy and stem cells?”

4.30-5pm

- Conclusion