

THE PROPOSERS

The International Ring14 Association (www.ring14.org) has promoted and financed the workshop. The Scientific Committee has made the event possible by developing the idea and the scientific program.

This committee is composed of:

- **Giovanni Neri**, MD Università Cattolica del S. Cuore, Rome, Italy.
- **Stylianos Antonarakis**, MD University of Geneva, Geneva, Switzerland.
- **Pietro De Camilli**, MD (father of a Ring 14 child), Yale University, New Haven, CT, USA.
- **Emilio Perucca**, MD University of Pavia, Pavia, Italy.

VENUE AND DATE

The workshop will take place in the characteristic Langhe district of Piedmont (Region of North West Italy). A land of wines - Barolo and Barbaresco - and of culinary excellence ranging from cheeses to nuts, including of course the white truffle of Alba. The countryside is dotted with castles, manors and towers, veritable storehouses of history and art, and the area is being proposed as a Unesco World Heritage site. Accommodation has been booked at the four-star Albergo dell'Agencia in Pollenzo, Bra in the Province of Cuneo (Italy) - www.albergoagenzia.it.



CONTACTS

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Under the auspices of:



Provincia di Como



UNIVERSITÀ
CATTOLICA
del Sacro Cuore

ring14

AIUTO E RICERCA PER I BAMBINI AFFETTI
DA MALATTIE GENETICHE RARE - ONLUS.

IMPEGNO, SOSTEGNO, FUTURO.

1st WORKSHOP

ON

RING14 SYNDROME

October 1 - 4, 2011

Pollenzo, Bra - Italy

www.ring14.org



THE WORKSHOP

The workshop is promoted by the **International Ring 14 Association**, based in Reggio Emilia, Italy. The purpose of the Association, founded in 2002, is to facilitate and stimulate scientific research on mutations of chromosome 14, which up to now have been largely unknown to researchers. The workshop represents the realisation of a dream: to arrange a time for meeting and study between researchers of international excellence on rare and hitherto unknown syndromes.

OBJECTIVES OF THE WORKSHOP

The workshop wants to stimulate research on Ring14 syndrome. The workshop will gather together a small number of scientists from different fields, with the aim of generating seminal ideas and new approaches. These scientists are expected to discuss and suggest strategies to identify the genes on chromosome 14 that play dominant roles in the manifestations of the syndrome, the function of these genes, and the mechanisms of disease that might be amenable to treatment.



PROGRAM

01 OCT - SATURDAY

Arrival of all invited speakers at Hotel in the morning/afternoon. Those who will arrive by plane will have a transfer to the Hotel.

17,00 Wellcome drink.

17,30- 18,00 Opening remarks by members of the Scientific Committee.

18,00 – 18,30 Presentation of the Association (Stefania Azzali).

18,30 – 19,00 The language's study on Ring 14: Prof.ssa D'Odorico Laura (Università Bicocca Milano - Italy).

20,00 Dinner in Hotel with Ring14's families and overnight.

02 OCT - SUNDAY

Breakfast at Hotel.

Full day meeting.

Chairperson: Giovanni Neri (Roma - Italy).

9,00-9,45 Marcella Zollino (Roma - Italy): What is the ring 14 syndrome: a study of 40 patients.

9,45-10,30 Leslie Biesecker (Bethesda, MD - USA): Genotype-phenotype correlations in chromosome syndromes.

10,30-11,15 Stylianos Antonarakis (Geneva - Switzerland): Trisomy 21: a model system for the study of chromosome syndromes.

11,15-11,45 Break.

11,45-12,30 Nancy Spinner (Philadelphia, PA - USA): The experience with the ring chromosome 20 syndrome.

12,30-13,15 Orsetta Zuffardi (Pavia - Italy): The FOXP1 gene and epilepsy.

13,15-14,30 Lunch in the Hotel.

Chairperson: Stylianos Antonarakis (Geneva - Switzerland).

14,30-15,15 Yann Herault (Strasbourg - France): Can animal models be built for a ring chromosome syndrome?

15,15-16,00 Anne Cherry (Boston, MA - USA): iPSC cells, a tool to study the neurobiology of ring 14 cells.

16,00-16,30 Break.

16,30-17,15 James Lupski (Houston, TX - USA): A strategy to identify relevant genes in a chromosome syndrome (video conference).

17,15-18,00 Murat Gunel (New Haven, CT - USA): Genome-wide studies and chromosome syndromes.

19,00 – 20,00 Visit to The “Banca del Vino”, located in the historical cellars of the Agenzia di Pollenzo. The Banca del Vino is host to a museum expanding of over two thousand square meters where you can transverse the Italian oenological landscape, stopping, region by region, tasting the wines that represent its history.

20,00 Dinner in the Hotel with The “Slow Food Presidia”, who protects small-scale local products which are at risk of disappearing. Overnight in the Hotel.

03 OCT MONDAY

Breakfast at Hotel and full day meeting.

Chairperson: Emilio Perucca (Pavia - Italy).

9,00-9,45 Renzo Guerrini (Firenze - Italy): Chromosomal disorders and epilepsies: pathogenetic mechanisms and phenotypic variation.

9,45-10,30 Giuseppe Gobbi (Bologna - Italy): Ring 14 - associated epilepsies: Seizure semeiology, EEG features and prognostic factors.

10,30-11,00 Break.

11,00-11,45 Maria Paola Canevini (Milano - Italy): Epilepsy in the ring 20 syndrome.

11,45-12,30 Steve White (Salt Lake City, UT - USA): How can we develop more effective antiepileptic drugs.

12,30 -14,30 Lunch in the Hotel.

Chairperson: Pietro De Camilli (New Haven, CT - USA) – co-chair Giovanni Neri (Roma - Italy).

14,30 – 15,00 The RING14 collection in the TGBN Galliera Biobank (Franca Dagna Bricarelli, Genova - Italy).

15,00 – 15,30 The RING14 clinical data base (Emanuela Ponzi, Roma - Italy).

15,30-17,30 General discussion and proposals for the future.

18,00 Departure from Hotel by bus for the visit to the “Village of Barolo”. Barolo is a small hilltop village, surrounded by vineyards that every day renew the emotion of the life, whether you are in the sunlight or in the secret dark of some cellar. Dinner in the famous restaurant “Marchesi di Barolo” and return at Hotel for overnight.

04 OCT TUESDAY

Breakfast at Hotel
Departure and transfers at airport.

