

## THE PROPOSERS:

- **Ring14 Italy Onlus Association** promoted and supported the organization workshop, in collaboration with **Ring14 USA Outreach**;
- The **Scientific Committee** of the Workshop was in charge of the scientific Program.

The members of the Committee are:

**Stylianos E. Antonarakis** (University of Geneva Medical School, Geneva, Switzerland) - Chair

**Nancy Spinner** (The Children's Hospital of Philadelphia, Philadelphia, USA)

**Alexandre Raymond** (University of Lausanne, Lausanne, Switzerland)

**Jeffrey Noebels** (Baylor College of Medicine, Houston, USA)

## PLACE AND DATE:

The workshop will take place in the picturesque Franciacorta, a splendid region of Lombardy, a stone's throw from Brescia and within easy reach of the rest of Europe. For thousands of years, nature has sculpted this ancient land, creating a breathtakingly varied landscape awash in color, which yields its most prized treasure, wine. Yet, its history has been molded by man, who has toiled for centuries to create an impressive cultural heritage. The Associazione Strada del Franciacorta was set up to promote and preserve this heritage.

Accommodations have been booked at the charming 5-star resort Relais et Chateaux L'Albereta, in the verdant hills of Franciacorta a few minutes' drive from Lake Iseo, an enchanting place off the beaten track. The ideal getaway for those seeking the pleasure of "some time-off", L'Albereta seduces its guests with refined proposals for haute cuisine and SPA of Espace Vitalité Henri Chenot wellness & medical center.

## L'ALBERETA RELAIS & CHATEAUX

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How to reach the Albereta Resort: Situated in the center of Lombardy, a mere half-an-hour drive from Brescia and Bergamo and less than an-hour from Milan Linate or Malpensa and other major cities of art in North Italy.

## CONTACTS:

### RING14 ITALY ONLUS ASSOCIATION

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with the collaboration and support of:



# 2<sup>nd</sup> WORKSHOP ON RING14 SYNDROME



*25-27 October 2014  
Erbusco (BS)  
Italy*

## BACKGROUND:

The workshop is promoted by Ring14 Italy Onlus Association, based in Reggio Emilia, Italy in collaboration with its sister organization Ring14 USA. The mission of the International Ring14 Association is to stimulate and foster scientific research on alterations of chromosome 14, which have been largely neglected by the scientific community so far.

The workshop on Ring 14 Syndrome, in its second edition this year, is a dream come true, that will gather together world-class experts in order to discuss mechanisms of chromosome 14 related syndromes with the ultimate goal of paving the way for a concrete therapy.

## AIM AND EXPECTED OUTCOME:

The primary aim of this workshop is to stimulate scientific research on Ring 14 syndrome. Towards this end, we will bring together excellent scientists working in different but relevant fields, in order to generate a stimulating and interactive environment where new research ideas can be developed and explored. Among the invited speakers, there will be some scientists who do not possess any specific expertise on Ring 14 syndrome. It is hoped they will contribute to our understanding of the Ring 14 Syndrome by bringing an original perspective to the discussion.

Given the topics that will be addressed and the expertise of the participants, the workshop should provide a focus on strategies to identify genes that play dominant roles in the manifestations of the syndrome. Furthermore, an examination of the functional features of these genes should lead a greater understanding of disease mechanisms that will ultimately be used in developing new potential treatments.

## MEETING AGENDA:

### 25 October 2014, Saturday

AM	Arrival, welcome drink and rooms allocation.
12.00-13.15	Light Lunch
15.15-15.25	Presentation of the Ring14 Association (S. Azzali & Y. DeWoody)
15.25-15.40	Giovanni Neri (Università Cattolica del Sacro Cuore, Rome, Italy) emeritus member of the Ring14 Scientific Committee
15.40-15.45	Opening Remarks by S. E. Antonarakis (University of Geneva Medical School, Geneva, Switzerland) - Chair of the Scientific Organizing Committee
15.45-19.00	<b>Session 1 - Ring14 syndrome: from rearrangement characterization to disease modelling. Chair: Alexandre Reymond (University of Lausanne, Lausanne, Switzerland)</b>
15.45-15.55	Alexandre Reymond (University of Lausanne, Lausanne, Switzerland): Introduction to the session
15.55-16.25	Orsetta Zuffardi (University of Pavia, Pavia, Italy): "Chromosome 14q distal deletions in linear and ring chromosomes: any breakpoint peculiarity?"
16.25-16.55	Nancy Spinner (The Children's Hospital of Philadelphia, Philadelphia, USA): "Overview of ring chromosomes: similarities and differences"
16.55-17.10	Coffee Break
17.10-17.25	Francesca Sofia (Advisor for Ring14 strategic management): "Clinical database of the Ring14 Association: state of the art"
17.25-17.55	Yann Héroult (IGBMC, Strasbourg, France): "Modeling Ring14 in mice"
17.55-18.25	Anthony Wynshaw-Boris (Case Western Reserve University, Cleveland, USA): "Chromosome therapy during reprogramming in patient-derived induced pluripotent stem cells"
18.25-18.40	Didem Demirbas Cakici (Boston Children's Hospital, Boston, USA): "Reprogramming Ring 14 Syndrome"
18.40-19.00	Final Discussion
20.00	Dinner

### 26 October 2014, Sunday

07.30-08.30	Buffet breakfast
08.55-11.45	<b>Session 2 - Effects of the Ring14 chromosome on genome organization, epigenetics and cell cycle control. Chair: Stylianos E. Antonarakis (University of Geneva Medical School, Geneva, Switzerland)</b>
08.55-09.00	Stylianos E. Antonarakis (University of Geneva Medical School, Geneva, Switzerland): Introduction to the session
09.00-09.30	Hendrik G. Stunnenberg (Nijmegen Centre for Molecular Life Sciences, Nijmegen, The Netherlands): "Epigenomics: functional indexing genomes in health and disease"
09.30-10.00	Phil Hieter (The University of British Columbia, Vancouver, Canada): "Chromosome Structure and Function: Lessons from Yeast"

10.00-10.15	Coffee Break
10.15-10.45	Stylianos E. Antonarakis (University of Geneva Medical School, Geneva, Switzerland): "Genome function with a few extra megabases"
10.45-11.15	Christian Bar (Spanish National Cancer Research Centre CNIO Madrid, Spain): "Telomeres and the origin of disease"
11.15-11.45	Final Discussion
12.00-13.00	Light Lunch
13.15-16.00	<b>Session 3 - Gene expression alterations and single cell transcriptome. Chair: Nancy Spinner (The Children's Hospital of Philadelphia, Philadelphia, USA)</b>
13.15-13.20	Nancy Spinner (The Children's Hospital of Philadelphia, Philadelphia, USA): Introduction to the session
13.20-13.50	Thomas Cremer (Ludwig-Maximilians-University in Munich, Germany): "Functional organization of the cell nucleus: experiments and model views"
15.50-14.20	Alexandre Reymond (University of Lausanne, Lausanne, Switzerland): "Genome structure, chromatin spatial organization and gene expression"
14.20-14.50	James H. Eberwine (University of Pennsylvania, Philadelphia, USA): "Single Cell Genomics - The role of Transcriptome Variability in Cellular Function"
14.50-15.20	Jason Mills (The Children's Hospital of Philadelphia, Philadelphia, USA): "Cellular models of ring chromosomes"
15.20-15.40	Final discussion
15.40-16.00	Coffee Break
16.00-19.45	<b>Session 4: Epilepsy session (genetics, and structural/functional MRI and brain alterations/mosaicism-somatic mutations). Chair: Jeffrey Noebels (Baylor College of Medicine, Houston, USA)</b>
16.00-16.30	Jeffrey Noebels MD, PhD (Baylor College of Medicine, Houston, USA): "Introduction to the session and Update on the biology of epilepsy genes"
16.30-17.00	Federico Vigevano, MD (Ospedale Pediatrico Bambino Gesù, Rome, Italy): "Clinical neurophysiology of epilepsy in Ring 14"
17.00-17.30	Jeff Golden (Harvard Medical School, Boston, USA): "Building a case for interneurons in epilepsy: Implications for Ring 14"
17.30-17.45	Final Discussion
17.45-19.45	Round table: All speakers are invited to discuss and design new avenues for research on Ring 14 syndrome. Chairs: Stylianos E. Antonarakis (University of Geneva Medical School, Geneva, Switzerland), Nancy Spinner (The Children's Hospital of Philadelphia, Philadelphia, USA), Alexandre Reymond (University of Lausanne, Lausanne, Switzerland), Jeffrey Noebels (Baylor College of Medicine, Houston, USA) and Pietro de Camilli (Yale University School of Medicine, New Haven, USA)
20.30	Dinner

### 27 October 2014, Monday

Buffet Breakfast and departure.

