

## BACKGROUND:

The workshop is promoted by the Ring14 International Association through the support of all its national organizations (i.e., Ring14 Italy, Ring14 USA, Ring14 France, Ring14 Spain, Ring14 Netherlands, and Ring14 Belgium). The purpose of the Association, founded in 2014, is to coordinate the national chapters as well as to facilitate and stimulate scientific research on rearrangements of chromosome 14, which up to now have been largely neglected.

This 3rd workshop represents the continuation of a dream: to bring together researchers of international excellence to discuss mechanisms underlying chromosome 14 related syndromes and to define research initiatives with the potential to ultimately lead the way to effective therapies.

## OBJECTIVES:

The objective of the meeting is to further stimulate research on Ring 14 syndrome. The Workshop will bring together a small number of scientists from different fields, with the aim of generating new ideas and developing new therapeutic approaches. Most invited speakers are not experts in Ring 14 syndrome. Rather, they are expected to share their expertise in different fields relevant to Ring14 syndrome to conceptualize strategies towards the identification of genes that play key roles in the manifestations of the syndrome, the functional characterization of these genes, and the mechanisms of disease that might be amenable to treatment.

## SETTING:

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. Nature has over thousands of years sculpted this ancient land, creating a breathtakingly varied landscape awash with color, which yields its most prized treasure, the wine.

## HOTEL VILLA FERNAROLI

Via Giuseppe Mazzini, 14 - 25086 Rezzato (BS)  
Tel: +39 030 2793223; Fax: +39 030 2594111  
Hotel info@villafenaroli.it

This charming 4 sup-star hotel is situated near the international airports of Milan Linate (110 km) and Milan Malpensa (150 km) and the regional Aeroporto di Brescia (15 km).

## Contacts:

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with the collaboration and support of:



# 3<sup>rd</sup> WORKSHOP ON RING14 SYNDROME



25-27 October

Franciacorta (BS)

Italy *ut labiqua.*

## 25 OCTOBER 2019, FRIDAY

**AM** Arrival, welcome drink and room allocation.

### Buffet lunch

### Introduction and overview of Ring14

- 14.30** Yssa DeWoody (Texas, USA), President of Ring14 International, "Our family's story; this foundation's story"
- 14.50** Giovanni Neri (Rome, IT), Chair of the Ring14 Scientific Advisory Board, "A clinical overview on Chromosome 14 syndromes"
- 15.10** Marco Crimi (Milan, IT), Scientific Coordinator Ring14 International, "An overview on research activities promoted by Ring14 International"
- 15.30** Coffee Break

### Session 1: Cellular modeling for Ring14 and other neurodevelopmental disorders. Chair: Giovanni Neri (SAB Ring14 International, Italy)

- 16.00** Anthony Wynshaw-Boris (University Hospitals Case Medical Center, Cleveland, USA), "Ring chromosome loss during iPSC reprogramming"
- 16.30** Jack Parent (University of Michigan, Ann Arbor, USA), "Modeling neurodevelopmental disorders using human pluripotent stem cells"
- 17.00** Jenny Hsieh (University of Texas, San Antonio, USA), "Human 3D brain organoid models of ARX-associated neurodevelopment disorders"
- 17.30** Robert Hunt (University of California, Irvine, USA), "Interneurons in neurodevelopmental disorders"
- 19:00** **Departure for wine cave visit and Dinner at restaurant Barboglio De Gaioncelli**  
<https://www.barbogliodegaioncelli.it/>



## 26 OCTOBER 2019, SATURDAY

### Buffet breakfast

### Session 2: Ring14 and other rare genetic epilepsies: genetics and treatments. Chair: Stylianos E. Antonarakis (University of Geneva, Switzerland)

- 09.00** Annamaria Vezzani (Mario Negri Institute, Milan, Italy), "A rat model of de novo status epilepticus to study long-term consequences of acute injury and seizures in the Ring14 Syndrome"
- 09.30** Nicola Specchio (Ospedale Pediatrico Bambin Gesù, Rome, Italy), "Ring 14 syndrome: clinical and neurophysiologic findings"
- 10.00** Rana Said (UT Southwestern Medical Center, Children's Health Dallas, USA), "Management approach to treatment of epilepsy associated with Ring Chromosome 14"
- 10.30** Coffee Break
- 11.00** Scott Baraban (UCSF, San Francisco, USA), "Using zebrafish to model rare genetic epilepsies"
- 11.30** Tracy Dixon-Salazar (LGS foundation, San Diego, USA), "Thinking about Ring 14 clinical trials: lessons and comparison from LGS"
- 12.00** Erin Heinzen (Columbia University, New York, USA), "Somatic mosaicism in epilepsy disorders"

### Buffet Lunch

### Session 3: Research tools on ring chromosome associated syndromes. Chair: Andrew DeWoody (Purdue University, USA)

- 14.00** Andrew DeWoody (Purdue University, USA), "Potential avenues for the study of Ring 14: a paternal perspective from an evolutionary biologist"
- 14.30** Peining Li (Yale University, USA), "Human ring chromosome registry: A practice resource for genetic diagnosis, clinical management and research collaboration"
- 15.00** Mariaclara Bonaglia (IRCCS E. MEDEA, Bosisio Parini, Lecco, Italy), "Emerging impact of 3D genome on ring chromosome-associated syndromes"

### Session 4: Pro-bono projects and EU-funding opportunities. Chair: Marco Crimi (Ring14 International, Italy)

- 16.00** Domenico Garozzo (CNR, Catania, Italy), "Serum N-Glycome studies in Ring14 patients"
- 16.20** Generoso Iannicello (Dantelabs, USA/Italy), "Human Whole Genome Sequencing: applications and results"
- 16.40** Berardo Rinaldi (University of Milan, Italy), "Ring14 syndrome: a gene-expression disorder?"
- 17.00** Julkowska Daria (INSERM, Paris, France), "European Joint Programme on Rare Diseases bringing opportunities to rare diseases community"
- 18.30** **Departure for Garda Lake for a short promenade and dinner at Restaurant QB Duepuntozero**  
<https://www.qbduepuntozero.com>

## 27 OCTOBER 2019, SUNDAY

### Buffet breakfast

- 8.00** Scientific Advisory Board meeting
- 9.00** **Round table: All speakers are invited to discuss and design new strategies for research on Ring14 and other chromosome 14 syndromes**  
Chair: Pietro De Camilli (Yale University, USA)
- 11.00** Yssa DeWoody, Farewell to speakers and funding opportunities
- 11.15** **End of Workshop for speakers and participants**
- 11.30** **Wrap-up meeting (restricted to Ring14 International & Scientific Advisory Board members) - led by Giovanni Neri**
- 12:30** **Buffet lunch and departures**

