

**PROF. GIOVANNI NERI**

**Director of the Institute of Genetic Medicine at the "Università Cattolica" of Rome**

**1° SCIENTIFIC INTERNATIONAL WORKSHOP**

**1) Prof. Neri, why did you join the Scientific Committee at RING14?**

I joined the Committee because of my general interest in chromosomal syndromes; ring chromosome syndromes are particularly "intriguing" in terms of molecular-clinical correlation. Moreover, I grew up in Reggio Emilia, so there is also a sentimental reason.

**2) What is your speciality and what kind of scientific activity do you carry out?**

I am a medical geneticist and my research primarily concerns the genetic causes of mental retardation and malformation syndromes. Currently I am also involved in clinical trials for the pharmacological treatment of mental retardation in fragile X syndrome.

**3) In all these years of history of RING 14, what advances have been made in the genetic study of chromosome 14 syndromes?**

Many case histories have been collected and on each patient was conducted an extensive molecular cytogenetic study on the one hand, and clinical study on the other, paying particular attention to the structural analysis of the ring, the physical and behavioral phenotype and epilepsy. Furthermore, an electronic database has been set up containing all the clinical and genetic information concerning each patient collected to date, as well as a biobank, which stores all the DNA samples and cell lines of many of the patients who belong to the Association.

**4) In 2009 the prestigious "American Journal of Medical Genetics" devoted a space to RING14. Can we define this as a first scientific milestone?**

It is indeed a milestone, which has helped to raise awareness of the syndrome and provided a stimulus for further studies.

**5) In October 2011 the first summit of chromosome 14 syndromes will commence. Who will participate? What are the objectives of the meeting and what will be the outcomes?**

In the Workshop scheduled for next October will participate a selected group of high-level international researchers who expertise in the field of cytogenetic, molecular genetics, clinical genetics and epilepsy in particular. The Workshop will be held in an informal environment to encourage discussion and promote further research in the field.

**6) What do you expect from this scientific brainstorming?**

I expect that the interest in the ring 14 syndrome will increase and will strengthen the collaborative relationship between the research groups, and will encourage new groups to engage in the study of the syndrome.