PROJECT ON THE GENETIC BASIS OF THE RING14 SYNDROME

Project Leader	Giovanni Neri
Host Institution	Istituto di Genetica Medica, Università Cattolica di Roma (Italy)
Duration	8 years
Start-up year	2004
Budget	€ 171.200
Status	Completed

Biomedical research on genetically triggered syndromes should always start from the study of molecular bases. In order to develop an effective therapy, it is fundamental to understand the role of genetic alterations and find the exact correlation between phenotype (set of all physical characteristics) and genotype (genetic makeup). Therefore, in the case of the Ring14 syndrome is important to correlate quantity and type of lost genetic material (deletions) or rearranged material (translocations or ring structures) with clinical symptoms.

It is logical to think that greater amounts of lost genetic material would lead to more severe symptoms, but it is equally important to understand the role of lost or damaged genes since they are fundamental in determining certain characteristics or traits. Genetic studies require a huge scientific, financial and organizational commitment on the part of Ring14 Association.

Since 2004, the genetic project has been, in fact, one of the main challenges for the Association, which relies on the valuable collaboration of the Institute of Medical Genetics at the Catholic University of Rome.

Initially, blood samples of children and parents of the Association were collected with the aim of performing necessary genetic examinations and analysis. Then, over the years, scientific research has continued these studies by providing additional clinical data, which has led to the publication of the following:

- Zollino M, Ponzi E, Gobbi G, Neri G. *The ring 14 syndrome*. Eur J Med Genet. 2012 May;55(5):374-80. doi: 10.1016/j.ejmg.2012.03.009. PubMed PMID: 22564756.

- Zollino M, Seminara L, Orteschi D, Gobbi G, Giovannini S, Della Giustina E, Frattini D, Scarano A, Neri G. *The ring 14 syndrome: clinical and molecular definition*. Am J Med Genet A. 2009 Jun;149A(6):1116-24. PubMed PMID: 19441122.

SCHOLARSHIPS AND DOCTORATES SUPPORTED BY RING 14 FOR THE PROMOTION OF GENETIC RESEARCH

- 2004: Scholarship devoted to the analysis of the first blood samples sent by Ring14 families.
- 2005/2007: Three-year PhD assigned to Dr. Laura Seminara (Catholic University of Rome) focused on the project "The Ring 14 syndrome: Clinical and Genetic Data". This was the first international study, conducted on 27 patients of the Association that used fluorescence in situ hybridization (FISH) and has allowed for the precise identification of DNA breakage and loss of genetic material chromosome.
- 2008-2009: Scholarship for Dr. Orteschi (Catholic University of Rome) who carried out a sophisticated analysis using the Microarray technique. This research has provided a more precise and detailed map of lost genes, leading to a correlation of genetic expression and the role of lost genetic material.
- 2010-2011: Scholarship for Dr. Ricciardi (Catholic University of Rome) for the study of RNA and therefore understand the function of lost genes in Ring14 syndrome.