Ring14 International grant to a Mario Negri Institute project to study epilepsy

Researcher Annamaria Vezzani from the Mario Negri Institute in Milan will study drug-resistant epilepsy with a grant received being the winner of an international call promoted by the "Ring14 International" association, set up with the aim of promoting research on syndromes related to chromosome 14 alterations.

Ring14 indicates an alteration of chromosome 14, which acquires a particular conformation, the typical ring shape, with a partial loss of genetic material of chromosome 14. This chromosomal abnormality can affect every cell or only a cell line, with the loss affecting a complete chromosome 14 and, in this case, the disease is called "monosomy 14". The damage is associated with a number of frequent signs and symptoms and results in a syndromic disease characterized by intellectual disability in addition to multiple disorders of the central nervous system, such as motor disorders, language disorders, microcephaly (i.e. redaction in brain size) and epilepsy.

The main project of Mario Negri of Milan will focus on identifying new therapeutic perspectives for drug-resistant epilepsy that affects patients with Ring14: epilepsy is in fact one of the main and well-established symptoms of this rare disease. The Ring14 International association has selected the project of the Italian researchers through a call for projects which allocated a grant of 50,000 euro to cover a 1-year research in this field which has not been adequately investigated so far.

"The call – said Marco Crimi, scientific coordinator of Ring14 International – aims at encouraging the progress of research in the field of chromosome 14 syndromes and at obtaining results to be

disseminated at congresses and published in scientific journals in order to obtain increasingly important grants".

"The ultimate goal – he continued – is to accelerate the path leading to the definition of therapeutic applications for children affected by these severe diseases; such an important objective can be reached only if the most deserving projects are awarded; and these projects can be identified thanks to the best review procedures: over the first two years of the call, we received the expressions of interest of researchers from all over the world".

The first two editions of the call granted funds to researchers submitting projects with high scientific value: the project by Anthony Wynshaw-Boris from Case Western Reserve University in Cleveland concerning the production of cell models of the syndrome and the project by Nancy Spinner from the Children Hospital of Philadelphia on the analysis of ring-shaped chromosomes 14 in human neuronal tissues.

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