

## **Aid for "Ring 14"**

### ***Association for Children with Chromosome Alteration***

It is a great challenge to talk about a mostly unknown disease for an association that constitutes an important scientific try. It is "Ring 14" founded in Reggio Emilia on 30<sup>th</sup> May 2002.

"This is an adventure that families and doctors have strongly pursued" explains Daniela Bruni, the vice-president, "the research activity and the awakening action start from the will to better understand a rare syndrome. Ring 14 is in fact an alteration of the chromosome 14 and it causes a set of multiple effects whose most common symptoms are the mental and motory retard, serious damages to the language ability, epilepsy and risk of infection at the respiratory system, and gastrointestinal disorders. It is a rare syndrome that counts about ten known and studied cases. This chromosome alteration could reveal to be more frequent, if the enquiries were held on a larger number of children."

"The essential thing", she goes on, "is to pay greatest attention to the early symptoms and signs of the patient. In case of precocious epilepsy or more or less evident deformations, it is important to do the chromosome test."

Daniela is Alessandro's mother, a 15-year-old boy affected by this rare genetic disease. Many have been the obstacles to overcome, even to get the right diagnosis. "The first signs of the disease came when Alessandro was a bit more than two years old, with the first epilepsy crisis. Since then hospitalizations and a lot of tests started to understand what kind of disease it was. When Alessandro was six, we met Dr. Giuseppe Gobbi. He found out Alessandro's genetic map, so, after years of doubts, we got the exact diagnosis. After we found an American medicine that could control the crisis. Alessandro hasn't had any for five years. Two years ago I got acquainted with four Italian mothers with the same problem. Then we felt the desire to create an association, in order to share this experience of ours and to support each other. It is very difficult to be mother/father of a child suffering from a rare disease: nobody can tell us exactly what will happen, what are the best remedies, what tests to do, how our child will grow up and what life quality he/she will have."

"Some of the aims of the association are: creating a databank, drawing-up a protocol for medical enquiries, political and scientific as well as social promotion of initiatives for diagnosis and research, fund raising, creating a net of doctors, health-operators and researchers, who have an interest in this rare pathology."

The Association central office is in Via Hugo 34, Reggio Emilia - Italy. Tel. 0039 0522 322607. The responsible person for Modena is Daniela Bruni, mobile phone 0039 349 5892062. To get more information about it visit the website [www.ring14.com](http://www.ring14.com), e-mail to [info@ring14.com](mailto:info@ring14.com).

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