New Ring 14 Research (2010-2011)  
Institute of Genetic Medicine Polyclinic A. Gemelli, Rome.

Genetic research on Ring 14 Syndrome has been planned, over the next two years, 2010-2011, along the following lines:

1) Analysis of expression of the *FOXG1* gene, candidate for epileptic encephalitis, through dosing of mRNA and the FOXG1 protein in patient lymphoblastoid cells with balanced translocation and phenotype compatible with proximal 14q deletion syndrome. Analysis of expression of the same gene on skin fibroblasts.

2) Expression analysis of the *FOXG1*, *NRL*, and *RPGR-IP1* genes, all candidates for retinal and macular degeneration, through dosage of mRNA and the corresponding protein in the a) lymphoblastoid cells and in b) skin fibroblasts of 5 patients with Ring 14, both complete and with deletion of the 14q terminal portion. A comparative evaluation will need to be carried out on lymphoblastoid cells and skin fibroblasts on subjects with normal karyotype, of similar age (this aspect will be conducted at the Institute of Genetic Medicine at Rome as well).

3) We will continue to study new cases through chromosomal examination of 100 cells, and through array CGH.

Lymphoblastoid cells are already available in the laboratory of the Institute of Medical Genetics, Catholic University of the Sacred Heart.

The skin fibroblast cultures must be prepared, so it is necessary to schedule a skin biopsy twice per month. The skin biopsy may be done at another location, but must be received at the Institute of Medical Genetics, Catholic University of Rome within the same day, or the skin biopsy may be performed at the Catholic University itself. The samples will be processed along with the completion of a consent form.

**Actually, each patient included in the study must provide:**

1) 7-8 ml of blood in HEPARIN  
2) a skin biopsy

The planned timeframe to complete both aspects is two years. These timeframes include both the performance of molecular research and any scientific publications.

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