PROJECT STUDY ON ABERRATIONS OF CHROMOSOME 14
PHASE 2 2007-2009: USE OF MICROARRAY –CGH TECHNOLOGY

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The "Microarray-CGH" technique aims to identify quantitative chromosomal abnormalities (partial deletions or duplications) smaller than the resolution limits of conventional chromosomal examination, and thus is called "cryptic".

Quantitative chromosomal abnormalities account for approximately 20-30% of cases of mental retardation. It should be noted that even in the presence of a chromosomal abnormality previously detected by standard chromosome examination, chromosomal rearrangements may be more complex. Multiple quantitative abnormalities may influence the final phenotype.
In the specific case of Ring 14 syndrome, there is evidence that some cases of rings are associated not only with partial loss of the terminal region of the long arm, but also to partial duplication of the proximal regions.

The technique is based on the following principles:

1) Extraction of DNA from peripheral blood of the subject in question

2) The DNA must first be hybridized with control DNA, in equimolecular amounts. The test DNA and control DNA are labeled with two different fluorophores, for example red for the test DNA and green for the control DNA. If the test DNA has no loss or duplication, the DNA hybridization between the two is complete, and the two DNAs cancel each other out in some way. If, however, the test DNA has a deletion, it exceeds, for that specific region, the control DNA; similarly, if the test DNA has a duplicate, it exceeds, for that region, the test DNA.

3) After mutual hybridization, the two DNAs are then hybridized with a series of molecular probes arranged on the same slide. The molecular probes are selected to cover the entire human genome, at a distance of 1 Mb or less, depending on the chosen degree of resolution. If the hybridization between the control DNA and the test DNA was complete (because there were no deletions or duplications), we obtain a continuous line, processed by special software; and if vice versa - the DNA in question is a deletion or duplication -we get a deviation in favor of the control DNA or test DNA, respectively. The deviation observed is limited to the region affected by the rearrangement, and simultaneously gives information on the extent of chromosomal defect, and the genes affected by the anomaly.

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