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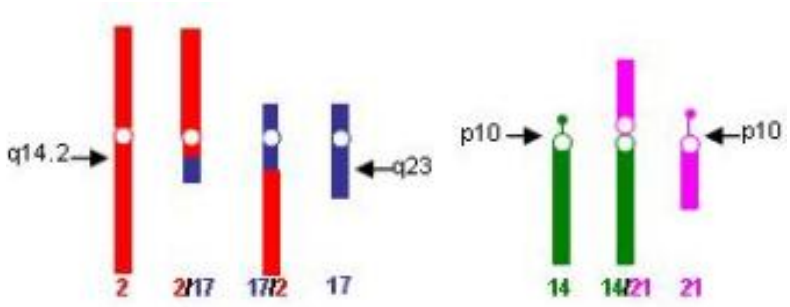
BARCELONA RECERCA I INNOVACIÓ

10/2012

Birth of the daughter of a carrier of double chomosomal translocations



In collaboration with a group from the Eugin clinic, formed by Dr. A. Obradors, Dr. R. Vidal, Dra. V. Vernaeve and Dr. O. Coll and with Dr. J. Obradors from the Consultori Obstètric i Ginecològic Josep Obradors, a group of researchers from the Unit of Cell Biology and Medical Genetics, formed by Dra. M. Rius, G. Daina, L. Ramos, Dr. J. del Rey, Dr. A. Obradors, Dr. J. Benet and Dra. J. Navarro, had achieved, for the first time with a fast Hibridació Genòmica Comparada (CGH) in the world, the birth of a child without a chromosomal alteration from a man that carried two chromosomal translocations, an alteration that minimized the successful transfer to the uterus of the mother.

Carriers of balanced chromosomal rearrangements, like translocations, have a risk of infertility or even the birth of children with chromosomal abnormalities due to the production of unbalanced gametes. In order to avoid this, a Preimplantation Genetic Diagnosis (PGD) is applied. In this case, a PGD analysing all the chromosomes was performed using a procedure, a comparative genomic hybridization (CGH) faster, developed in the Unit Cell Biology and Medical Genetics, Department of Cell Biology, Physiology and Immunology.

The PGD allows for selection and transfer to the maternal uterus, the embryos chromosomally normal or balanced detected to achieve healthy offspring. Conventionally, in PGD for chromosomal rearrangements, a serios of different techniques are applied in order to detect those chromosome imbalances derived exclusively from the particular reorganization, allowing for the discriminate between normal or balanced embryos from the ones unbalanced, but without

any analysis from the rest of the chromosome complement. With this procedure the European Society of Human Reproduction and Embryology that is collected that between 16% and 13% of embryos transferred achieved pregnancy showing heartbeat.

One reason for this low rate of implantation of embryos transferred from seems to be due to the presence also of chromosome abnormalities are not related to the chromosomes involved in the reorganization, which would also unfeasible.

In this case, alternatively to the up now, and in order to study simultaneously not only the chromosomes involved in the reorganization but also the rest of the chromosomes at the time, we applied the faster CGH. This method, which uses software MetaSystems (Izasa), allows for the simultaneously analysis of all chromosomes (chromosomes 1-22 and X and Y chromosomes) from the embryos. It also has the advantage that the results are achieved on four days which allows the transfer of the selected embryos to the maternal uterus without needs for freezing them.

The proceduers needed to select embryos not only chromosomally normal or balanced for one or both reorganizations but also chromosomally normal for the rest of the chromosomes, were performed in collaboration with gynaecologists and embryologists of the Clínica Eugin in which the whole Assisted Reproduction Technique was performed.

A total of 10 out of 12 embryos analyzed were evolved (83.3%) and were successfully diagnosed cytogenetically. Only one of them (10%) had no chromosomal imbalances (it was or normal or balanced for both chromosomes reorganization from the father moreover than euploide for the rest of the chromosomes). But the remaining (9/10) were unbalanced for some of the reorganization or both.

The embryo with no chromosomal imbalances were also morphologically very good quality and was transferred to the uterus four days after and resulted in the pregnancy and subsequent birth without any chromosomical alteration related to paternal translocations.

These results were included in a work that was published in the journal *Fertility and Sterility*. We have dedicated this work to the memory of the UAB Professor, Dr. Josep Egozcue Cuixart, pioneer of Human Cytogenetics of Reproductive Biology in Spain, an excellent scientist and teacher in addition to a very good friend.

PGD using the faster CGH is a useful tool to select embryos free of chromosomal abnormalities in any of the 23 chromosomes moreover than it can makes possible to increase the rate of introduction of PGD for family candidates to be carriers of chromosomal rearrangements.

Joaquima Navarro
Joaquima.navarro@uab.cat

References

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