First babies born from genetic screening study

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Two women taking part in the world's first controlled study of a comprehensive genetic screening test before IVF have given birth to healthy babies. The babies, twin girls born in Germany in June and a singleton boy born in Italy in September, are the first deliveries in a pilot study of comparative genomic hybridisation (CGH) by microarray, a new method of screening oocytes for IVF for a full range of chromosomal disorders.

Dr Cristina Magli, embryologist at the SISMER Centre in Bologna, one of the two centres taking part in the trial, said: " All the babies and their mothers are doing very well in terms of weight and overall developmental performance."

The births, as well as several ongoing pregnancies in the study group, are the final stage in the "proof of principle" that the screening of oocytes and embryos before transfer in IVF can increase birth rates; both these pregnancies were derived from oocytes whose complete chromosomal status had been assessed by microarray CGH.

The study, which was conducted in Bologna, Italy, and Bonn, Germany, was designed and organised by a task force of ESHRE to determine the clinical value of CGH as a non-subjective means of genetic screening before embryo transfer.

"We have learnt from more than 30 years of IVF that many of the embryos we transfer have chromosome abnormalities," explains

ESHRE's chairman Luca Gianaroli. "Indeed, it's still the case that two out of every three embryos we transfer fail to implant as a pregnancy, many of them because of these abnormalities.

"The whole world of IVF has been trying to find an effective way of screening for these abnormalities for more than a decade, but results so far have been disappointing with the technology available. Now we have a new technology in array CGH and our hopes are that this will finally provide a reliable means of assessing the chromosomal status of the embryos we transfer."

The microarray CGH technique as evaluated in the ESHRE study has several advantages over other methods:

- CGH tests all 23 pairs of chromosomes in a cell, and not just a limited number (as in former methods)
- The cell tested (known as the polar body) is taken from an oocyte at fertilisation, and so does not require biopsy of a cell from a developing embryo for its analysis
- Earlier chromosome tests were on cells biopsied from growing embryos and did not necessarily reflect the total status of the embryo (because of chromosome "mosaicism"); polar body analysis removes this potential problem
- Other CGH tests on biopsies from five-day-old embryos require several days to deliver complete results - and thus require the freeze-storage of the embryo before it can be transferred; polar body CGH can be done in real time and does not require freezing

At the everyday clinical level, polar body CGH is likely to have two

more important consequences: first, because the analysis is performed on oocytes and not on embryos, countries like Germany which outlaw embryo analysis and freezing will now have at their disposal a reliable method of preimplantation genetic screening; and second, because the chromosomal status of the transferred embryo can be accurately predicted (with no more than a 10 per cent error rate as found in the ESHRE study), the reduction of multiple pregnancies in IVF by single embryo transfer will become more attractive.

In the short term, the IVF patients most likely to benefit from preimplantation screening by polar body CGH are those of an older maternal age (over 37 years), those with a record of unsuccessful IVF, and those with a history of miscarriage; all these conditions are associated with a higher than average rate of embryonic chromosomal abnormality.

"The study has already caused huge interest in the scientific and clinical community," says Dr Magli, "and we are very proud to announce these results. It is the first time that a scientific society like ESHRE has organised a study to determine the clinical value of a technique which could prove a revolution in IVF."

The next step for ESHRE will be to upgrade the pilot study into a large-scale international clinical trial, which is planned to start in 2011.

Provided by European Society of Human Reproduction and Embryology

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