MedicalXpress

Karyomapping offers new way of detecting genetic conditions in IVF embryos

January 9 2015

New research at the University of Kent has identified karyomapping as a viable and cost-effective method of detecting a wide range of genetic diseases in IVF embryos.

The current method, which involves the use of preimplantation genetic diagnosis (PGD) for monogenic disorders (resulting from a single defective gene) has long been recognised as expensive, time-consuming and requires the tailoring of a specific test for each couple and/or disorder.

The research, led by Professor Darren Griffin of the University's School of Biosciences, in collaboration with researchers at Illumina, Inc. Cambridge, showed that karyomapping could be used, simultaneously, as a new approach to PGD for monogenic disorders with the potential for the detection of chromosomal disorders.

The new research is described in a paper published in December 2014, titled Karyomapping - a comprehensive means of simultaneous monogenic and cytogenetic PGD: Comparison with standard approaches in real time for Marfan syndrome, published by the *Journal of Assisted Reproduction and Genetics* in December 2014.

The paper also describes how in one clinical case study, a male partner was affected with Marfan syndrome, an autosomal dominant disease affecting the connective tissue which can lead to heart (aorta) and/or visual (retina) problems. Single cells from IVF embryos were biopsied

and analysed using standard PGD approaches including minisequencing for the Marfan mutation and analysis of three informative linked markers (work performed by the Dagan Wells laboratory at the University of Oxford and Reprogenetics UK).

Karyomapping was used to confirm the diagnosis utilizing a rapid 24-hour protocol enabling the researchers to perform karyomapping in a clinically applicable setting.

In a second clinical study, published earlier in 2014, karyomapping was used to confirm a PGD case detecting both chromosomal count (euploidy/aneuploidy) as well as a monogenic disorder (Smith-Lemli-Opitz (SLO) syndrome) simultaneously. In this study, the family underwent PGD with simultaneous diagnosis of both SLO status and chromosome constitution using standard approaches. Again, the diagnosis was confirmed simultaneously by karyomapping. (Live birth after PGD with confirmation by a comprehensive approach (karyomapping) for simultaneous detection of monogenic and chromosomal disorders. Natesan et al, 2014).

Both clinical cases led to the birth of a healthy child, unaffected by Marfan syndrome and SLO respectively.

More information: The paper Karyomapping - a comprehensive means of simultaneous monogenic and cytogenetic PGD: Comparison with standard approaches in real time for Marfan syndrome, is available at: link.springer.com/article/10.1 … 07/s10815-014-0405-y

Provided by University of Kent