

The London Bridge Fertility, Gynaecology and Genetics Centre and Virginia-based Genetics & IVF Institute Announce Universal Genetic Test

A team of UK and US researchers announced today that they can soon begin testing embryos for almost 15,000 inherited genetic diseases, using a new test called Karyomapping. Until now, it has been possible to test for only about 350 of the 15,000 diseases. Karyomapping was developed by The London Bridge Fertility, Gynaecology and Genetics Centre (Bridge), in collaboration with the Genetics & IVF Institute, Fairfax, VA. They plan to offer clinical trials of Karyomapping to couples seeking fertility treatment later this year.

The researchers who developed the test, led by Professor Alan Handyside, told a news briefing at the 25th annual meeting of the European Society of Human Reproduction and Embryology in Amsterdam that they have successfully screened cells taken from embryos that were known to have or carry cystic fibrosis.

The researchers from Bridge and Genetics & IVF Institute have proved that Karyomapping is capable of not only detecting diseases caused by a specific gene mutation, in this case cystic fibrosis, but that it is also capable of simultaneously detecting aneuploidy (an abnormal number of any of the 23 pairs of chromosome). This is the first time they have been able to demonstrate that the test can work in cells taken from embryos that have already been diagnosed with the cystic fibrosis gene mutation using conventional preimplantation genetic diagnosis (PGD).

To employ the test, a single cell (blastomere) is removed from a three day old embryo and, using the new Karyomapping technique, the cell can be tested to determine whether an inherited disease is present. The technique maps all of the embryo's chromosomes and can check virtually any gene. Karyomapping can also be done on later-stage embryos where more than one cell can safely be removed from the embryo.

Prior to the embryo biopsy, DNA samples are obtained through cheek swabs from the parents and a close relative, (usually a child who has the disease).

The information from the DNA tests is compared with DNA from the embryo to map how its chromosomes are constructed from genetic material inherited from four grandparents. The "family tree" or genetic map can determine whether the embryo has inherited chunks of chromosome that contain any faulty gene. Only embryos free of the disease would be transferred to a woman's uterus.

With recent developments in embryo freezing, Karyomapping could be used in a step-wise fashion where only one embryo is tested at a time until a genetically normal/chromosomally normal embryo is located and used for elective single embryo transfer. This step-wise use of the technique would reduce the cost of the test greatly and should still allow for high implantation and pregnancy rates while limiting the number of embryos that need to be transferred to achieve pregnancy. Elective single embryo transfer has been shown to reduce the likelihood of multiples in each pregnancy.