

New developments in reproductive medicine

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Three out of ten women who undergo polar body diagnosis go on to have a child. The extensive technique of polar body analysis (PBA) is described by researchers in reproductive medicine at Lübeck, Germany, in an article in the current edition of *Deutsches Ärtzeblatt International*, in which they present three successful cases and one failure.

Polar bodies are by-products of normal cell division (meiosis) of an oocyte—a woman's egg cell—on its way to maturation. For people with monogenic diseases, which are caused by gene defects, polar body analysis can give an indication of whether the healthy version of the gene is present in the egg before fertilization takes place. Monogenic diseases, such as cystic fibrosis, are caused by an altera-tion of one gene; this makes them different from polygenic diseases, such as diabetes mellitus, for example, which are caused by a combination of defects in several genes.

In the cases of four women who were carriers of a monogenic disease, their own egg cells containing the intact version of the gene concerned were fertilized, allowed to develop to the embryonic stage, and then transferred into the womb. Three of the women had a successful pregnancy and gave birth to a healthy child. One woman failed to become pregnant, even after a further round of PBA fol-lowed by embryo transfer, and decided not to continue the treatment. These four cases are among nine couples tested using PBA at Lübeck by Georg Griesinger and his co-authors because of a known risk that the mother might transmit a monogenic disease.



In Germany PBA is an alternative to preimplantation diagnosis (PID), which depending on the result can lead to termination of pregnancy with all the associated psychological and physical stresses. The disadvantages of PBA are: hereditary diseases transmitted through the father cannot be diagnosed, and in the case of autosomal recessive diseases egg cells that would have resulted in the birth of healthy heterozygous carriers are discarded. In Germany, PBA for monogenic diseases is at present performed at only two centers, in Lübeck and Regensburg.

More information: Dtsch Arztebl Int 2009; 106(33): 533-8

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