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Preimplantation genetic testing for aneuploidy: New improvements with non-invasive liquid biopsy technique

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Preimplantation genetic testing for aneuploidy (PGTA) was originally performed by analyzing the first and the second polar body. However, it has later been increasingly performed by sampling trophectoderm (TE) cells from blastocysts. Recently, there is increasing concern about the reliability of this technique which has actually never been tested sufficiently in animal models and human preclinical studies. The main problems of PGTA using TE biopsy can be resumed as follows: (1) The frequency of aneuploid TE cells does not necessarily reflect that in the inner cell mass (ICM) which will give rise to the future fetus, (2) the distribution of euploid and aneuploid TE cells is not random but rather clonal, making it impossible to obtain reliable information about the frequency of aneuploidy in the whole embryo, and (3) the removal of TE cells is inherently traumatic, can decrease embryo implantation potential and produce long-term effects on the offspring health. Since, in many cases, PGTA is performed in older women, with only few and relatively fragile embryos, the technique based on TE biopsy can lead to an irreparable damage due to accidental embryo destruction or voluntary destruction of viable embryos deemed aneuploid because of a false positive PGTA result. By contrast, PGTA using non-invasive liquid biopsy is based on analysis of cell-free DNA released both from TE and ICM cells to culture medium, thus allowing a more objective ploidy evaluation of the whole embryo. Here I present the latest data obtained by comparing ploidy evaluation results obtained from cell-free DNA analysis with those obtained by analysis of DNA obtained from whole embryos donated for research from consenting patients. These results show clearly the superiority of non-invasive PGTA based on liquid biopsy (cell-free DNA) from spent culture media over the conventional TE biopsy, with a considerable reduction of interpretation errors.

Speaker Biography

Jan Tesarik obtained his MD degree in 1979 and PhD in 1982. From 1989 he worked at the Americaan Hospital of Paris and achieved the world's first childbirths after round spermatid injection (ROSI) into oocyte cytoplasm. In 1998 he achieved, in Istanbul, the world's first childbirth after oocyte fertilization with spermatids obtained by in vitro spermatogenesis. He developed an original technique for nuclear transfer in mature human oocytes (Rome, 2000) and achieved the first fertilizable human "artificial oocytes" reconstructed from somatic cell nuclei and donor ooplasts (Sao Paulo, 2001). He described beneficial effects of growth hormone on oocyte quality in women of >40 years old. He is author of >400 scientific publications, including 307 highly influential publications listed in Semantic Scholar. At present he is Director of MARGen (Molecular Asssisted Reproduction and Genetics) Clinic in Granada (Spain) and coordinates different research projects carried out at the University of Granada.

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