# FETO-PLACENTAL DISCREPANCIES (FPD) IN PRODUCTS OF CONCEPTION (POCS) SHOW THAT ANEUPLOIDY IS NOT RESTRICTED TO PLACENTAL CELLS

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**Study question (25 words)**

In feto-placental discrepancies, is aneuploidy restricted to the placental cells?

**Summary answer (25 words):**

Hysteroembryoscopy allow us to detect accurately FPD (2%), with the presence of the aneuploid cell line in the fetal tissue.

**What is known already: (100 words)**

FPD are due the presence of two different cell lines in a pregnancy, usually presenting the fetus a normal karyotype and the placenta one abnormal, this is known as mosaicism confined to placenta (MCP). Despite MCP is described in normal evolution pregnancies, chromosomal abnormality present in placenta could lead a poor perinatal outcome even ending in an EPL. Nevertheless, if this abnormality is located in the fetus, the perinatal outcome even is worse, furthermore a prenatal non-invasive could lead to a false negative.

**Study design, size, duration (75 words):**

The objective of the study was to infer the real incidence of fetoplacental discrepancies in EPL analyzing samples obtained by hysteroembryoscopy from both, embryo and throphoblast tissues.

A total of 46 first trimester miscarriages in which at least two types of tissues were successfully ((fetal and extraembryonic) collected by hysteroembryoscopy were included in this study (January 2014- September 2016).

Hysteroembryoscopy allowed to accurately differentiate the tissues and to decrease the risk of maternal cell contamination.

**Participants/materials, setting, methods: (75 words)**

Specimens were collected in sterile tubes containing saline solution. Direct DNA extraction was performed without the previous cell culture required for classical cytogenetic studies. Chromosome status was determined by genetic molecular methods genome hybridization (CGH array) (Illumina, San Diego, USA) or Next Generation sequencing (NGS) (Thermofisher, MA USA). STRs analysis of maternal and specimen DNA was included in results showing female gender to fully discard MCC (AmpFlSTR Identifiler Plus (Applied Biosystems, CA, USA).

**Main results and the role of chance (200 words):**

In 45 of the 46 cases, the results were concordant between trophoblastic tissue and its corresponding embryo tissue. In only one case a discrepancy was reported, trophoblast with an euploid result (46,XX) and fetal sample with an aneuploid result (47,XY,+4), without maternal contamination. This case corresponded to a 30 years old patient with recurrent pregnancy loss that underwent preimplantation genetic diagnosis by aCGH cycle with transfer of an euploid embryo on day-3.

**Limitations, reasons for caution: (50 words)**

Limitations for the present study are the number of samples included and the fact that not the full sample of tissue could be analyzed.

**Wider implications of the findings: (50 words)**

In FPD, abnormalities are frequently confined to placenta. However, aneuploid cells can be also be preferentially located in fetal tissue. Therefore, the transference of mosaic embryos could increase the risk of poor perinatal pregnancies and abnormal fetus since abnormal cells are not always derived to placental tissue.

**Study funding/competing interest(s): optional**

**Trial registration number: (25 words)**