

## **Aneuploidy Screening in Blastocoel Fluid During Next Generation Sequencing (NGS)-Based Preimplantation Genetic Diagnosis (PGD)**

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**OBJECTIVE:** Recent study has demonstrated that the amount of present in blastocoel fluid DNA is insufficient for preimplantation genetic screening using array comparative genome hybridization. Our intention was to verify this results using more accurate technique of next generation sequencing.

**DESIGN:** This preliminary study included results from analysis of 8 embryos using next generation sequencing performed between January and February 2013 at Fertility Centre in Bialystok, Poland. Aneuploidy screening was performed in blastocoel fluid obtained from blastocyst stage embryos biopsied on day-5 of culture. Analysis of trophectoderm served as control.

**MATERIALS AND METHODS:** In total 8 embryos obtained from 2 patients were analysed. Blastocoel fluids from blastocyst stage embryos were screened for aneuploidies with next generation sequencing Ion Torren Suit Software. The study is ongoing and we continue to add samples.

**RESULTS:** Blastocoel analysis results don't show correspondence with results of trophectoderm analysis obtained from the same embryo. However, thresholds for gain or loss of chromosome were calculated for trophectoderm not for blastocoel fluid. It is possible that analysis based on thresholds recalculated for mix of inner cell mass and trophectoderm DNA content, which occur in blastocoel could provide more reliable results.

**CONCLUSIONS:** Blastocoel fluid analysis results only partially correspond with those obtained from trophectoderm, suggesting that recalculation of thresholds for chromosome specific DNA amount could result in better correspondence. If confirmed, testing of blastocoel fluid may become a new, less invasive method of preimplantation genetic diagnosis.