Detection of balanced translocations in Preimplantation Genetic Screening by means of next-generation sequencing: a 24-chromosome aneuploidy detection of 100 embryos

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Context: PGS involves the genetic study of the embryo’s genome to determine their chromosomal arrangement and in this way, selecting euploid embryos for implantation, increasing pregnancy success rates in IVF. Objective: Evaluation in terms of PGS for aneuploidy detection, how the NGS platform Ion-Torrent(TM) along with low-pass whole genome sequencing, provides the ability to evaluate many biopsied trophectoderm cells in order to confirm which euploid embryos were ready-to-transfer in an IVF cycle. Methods: Double-blinded genetic testing with both NGS and aCGH methods of 100 WGA samples amplified by PicoPlex® technique from trophectoderm cells. Patients: A total of 29 female patients (median age 40±5.57 years) were recruited in 30 clinical IVF cycles. Interventions: The ovocytes were inseminated using ICSI following an in vitro procedure. Resulting embryos were cultured until day 5 and trophectoderm biopsy was carried out on 100 blastocysts. Main Outcome Measure: NGS is rapid, highly accurate, robust, sensitive, easily reproducible, and a cost-effective method for detection of chromosomal aberrations that have a significant impact on health or embryo viability. Results: The comparison of PGS results by NGS to well-established aCGH have been concluded with 97.9% (95% CI: 92.6%-99.7%) diagnosis correspondence by both techniques. Both aneuploidy detection and a precise identification of balanced translocations were assessed. The discordance in two embryos was due to a more accurate and reliable screening solution by NGS. Conclusions: This validation study demonstrates the clinical application of this aneuploidy detection method in PGS by NGS in contrast to well-established aCGH. To be fully described in the Program? of the II Satellite? Symposia "New Developments in Genetics". For questions please contact IVF Spain Foundation: c.bezos@ivf.spain.com and Prof. Genazzani.