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AB094. Efficacy of combined preimplantation genetic diagnosis (PGD) and preimplantation genetic screening (PGS)

Part 4: Oral/poster

AB094. Efficacy of combined preimplantation genetic diagnosis (PGD) and preimplantation genetic screening (PGS) cycles—early results

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Abstract: Preimplantation genetic diagnosis (PGD) using PCR allows couples where one or both carry a hereditary single gene disorder to avoid having a child with that disorder. It can also be an effective therapeutic tool in curing an existing affected sibling through tissue matched cord blood stem cell transplant. However early preimplantation embryos have significant levels of chromosomal aneuploidy increasing with maternal age. Recent PGS technologies such as comparative genome hybridization (CGH) allow screening of all 24 chromosomes in the early embryo, allowing selective single embryo transfer (eSET) with significantly increased IVF implantation rates and significantly decreased miscarriage rates. We discuss early results on the efficacy of using PGD-PCR in combination with PGS-CGH (combined cycle) in couples who present for PGD for hereditary single gene disorders. PGD-PCR patients have a family specific test established, with the test components multiplexed and checked for reliability on single maternal cumulus cells. Patients having combined cycle had the individual test components checked on existing whole genome amplification (WGA) products and, if unreliable, reverted back to a standard PGD-PCR test/cycle only. Couples had an ovarian stimulation cycle, harvested eggs were fertilized using intracytoplasmic sperm injection (ICSI), and resultant normally fertilized embryos cultured to day 5 and day 6 blastocyst stage.

Suitable blastocysts were biopsied with assistance of a near-infra-red laser. The 1-6 cells obtained had their DNA extracted and either PCR amplified using the established multiplexed PGD-PCR test (PGD-PCR cycle) or WGA amplified (combined cycle). From 2007-2014, 109 couples presented for PGD-PCR for 16 different familial single gene disorders, predominantly beta-thalassemia (61/109) or alpha-thalassemia (25/109). In 2012 we introduced PGS-CGH for 24 chromosome screening of infertility couples, and soon after offered PGD-PCR patients the option of a combined PGS-CGH and PGD-PCR cycle; to date 19 patients had requested the combined cycle. For PGD-PCR only, 97 patients had 154 cycles with 85 embryo transfers (114 embryos). 57/85 (67%) were clinically pregnant with an implantation rate of 50%. For requested combined cycles, 5/19 patients (all alpha-thalassemia) failed the WGA check and reverted to PGD-PCR test/cycle only. 11/14 had 14 cycles with 8/14 cycles freeze-all (with no transfers to date) and 4 embryo transfers (5 embryos). 4/4 (100%) were clinically pregnant with an implantation rate of 80%. Early results, while low numbers, indicate offering patients presenting with a hereditary single gene disorder the option of having all 24 chromosomes screened prior to implantation may significantly increase their chance of a healthy pregnancy.

Keywords: Preimplantation genetic diagnosis (PGD); preimplantation genetic screening (PGS); array comparative genome hybridization (aCGH)

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