Al collegio docenti del Dottorato in Medicina Molecolare

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Introduction

Implantation is a developmental process that involves an intimate "cross-talk" between the embryo and uterus. Although there is increasing knowledge in the implantation process, the percentage of live birth rates in assisted fertilization cycles is around 30%. Embryos have been routinely selected for transfer during IVF on the basis of their morphology. However, morphological evaluation alone has not proved to be a very efficient tool for embryo selection; this is mainly because morphology does not necessarily correlate with the chromosomal status of the selected embryos. A significant proportion of infertility cases are due to genetic defects. Male infertility accounts for 50% of infertility cases; assessment of male infertility traditionally has depended on the analysis of a semen sample according to WHO guidelines; this analysis is based on a visual estimation of sperm number, motility and morphology as measured by light microscopy and is difficult to perform reliably. Male infertility is associated with impaired embryo development, higher miscarriage rates and increased risk of pregnancy loss, then testing is strongly recommended before a couple undergoes assisted reproduction in order to prevent the possible inheritance of the genetic lesion to the next generation. Chromosomal alterations, inversions, translocations, Y chromosome microdeletions, and gene mutations are the main genetic variants causing male infertility.

Methods

Next-generation sequencing (NGS) techniques permits the simultaneous interrogation of multiple disease-causing variants in many genes and detection of very different types of variants by using a single test in combination with multiple bioinformatics algorithms to process these data.

Results

In this year my work focused on bibliographic research and on the construction of the genetic panel to be used. We developed a NGS gene panel consisting of about 60 genes related to male infertility including 5′ and 3′ UTR for each gene, exons, and selected introns. The panel will simultaneously analyze the most widespread genetic factors that cause male infertility, such as Klinefelter syndrome, cryptorchidism, hypogonadism and genes with unsuccessful spermatogenic binding.
For patients with male infertility who show severe oligospermia or azoospermia the genetic panel could help explain the etiology and guide treatment.

Abstracts e partecipazione a congressi e corsi:

- Corso teorico-pratico sulla riproduzione assistita: Micromanipolazione e microambiente, 13-14 Novembre 2018, Napoli;
- Symposium 9.baby Bologna 25-27 Ottobre 2018;
- Best Practices in PMA, 8-9 Febbraio, Firenze;

Pubblicazioni scientifiche

- Laura Gambera, Anita Stendardi, Camilla Ghelardi, Benedetta Fineschi, Rosamaria Aini. *Effects of antioxidant treatment on seminal parameters in patients undergoing in vitro fertilization.* Archivio Italiano di Urologia e Andrologia 2019; 91,3: Submitted 2019, Accepted 2019.
- Laura Gambera, Francesca Vellucci, Anita Stendardi, Benedetta Fineschi, Silvana Pizzasegale, Camilla Ghelardi, Rosamaria Aini. *3 versus 6 oocytes in oocyte donors programs in Italy.* Reproductive BioMedicine Online, Submitted 2019