

CONSENT FOR PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

This consent form may contain words that you do not understand. Please ask the study doctor or the study staff to explain any words or information that you do not clearly understand.

You will receive a copy of this consent form for your records.

DESCRIPTION/PURPOSE

Preimplantation genetic diagnosis (PGD) is the chromosomal analysis of your eggs or embryos that may result from your in-vitro fertilization procedure. Genetic errors, such as changes in chromosome numbers, or aneuploidy, and changes in chromosome configuration will be studied. Aneuploid embryos are those with either a missing chromosome (monosomy) or an extra chromosome (trisomy). Aneuploidy occurs more frequently in eggs and embryos in women over 34 years of age. Changes in chromosome configuration include translocations. Couples in whom one or both partners have a known translocation can benefit. Translocation testing can determine the presence or absence of a certain chromosomal disorder, but cannot detect genetic disease nor predict congenital malformation. The study doctors and scientists at RBA hope to determine the genetic health of your eggs or embryos prior to transfer. The purpose is to select and replace only those embryos that appear to be chromosomally normal, so that there will be a reduced probability of losing the pregnancy or carrying a chromosomally abnormal baby to term.

The purpose of this procedure is to select and replace only embryos which do not have certain known chromosomal abnormalities.

OVERALL RATIONALE FOR UNDERTAKING THE STUDY

Chromosomes are the elements within every cell of your body that contain genetic information. Chromosomes are string-like structures found in the center of the cell, the nucleus. Inherited information is housed on the chromosomes. The traits are located in the genes that make up the chromosomes. Aneuploidies, or errors in development, occur more frequently in eggs and embryos in women over 34 years of age. Normally, there are 23 identical pairs in each cell, with a total of 46 chromosomes. Each patient provides 23 chromosomes, but in some children there is an extra chromosome: this is called trisomy. The most well-known trisomy is trisomy 21, also called Down's syndrome. Trisomic embryos usually do not implant, but if they do, this may lead to affected children. These extra chromosomes are usually formed during the final stages of egg ripening. The chromosomes mostly affected are chromosome numbers 13, 15, 16, 17, 18, 21, and 22.

A translocation is a change in chromosome configuration in which chromosomes are attached to each other or pieces of different chromosomes have been interchanged. An individual with a translocation is unaffected if there is no extra or missing chromosome material and if the break in the chromosome did not disrupt gene function. If there is no additional or missing chromosome material, the translocation is considered to be "balanced". A translocation is "unbalanced" if there is extra or missing material. Individuals with balanced translocations typically have no medical issues though some do have fertility problems such as reduced fertility. The concern regarding

