Comprehensive Chromosomal Screening (CCS): Information Sheet

OLIVE
fertility centre

Comprehensive Chromosomal Screening (CCS)

Chromosomal screening (CCS), otherwise known as pre-implantation genetic screening (PGS), is a diagnostic test that allows for the testing of fertilized eggs (embryos) to see whether they contain the appropriate amount of chromosomal (genetic) material.

Humans have 23 pairs of chromosomes, making 46 in total. When there is either more or less than 46 chromosomes the embryo is considered abnormal. Embryos with an abnormal amount of chromosomal material will not implant in the uterus, will implant and result in a miscarriage, or will implant and result in a pregnancy with an abnormal amount of chromosomes. An example of a condition with too much chromosome material is Down syndrome, where there is an entire extra chromosome 21 present. An example of too little chromosome material is Turner syndrome, where females have only one X chromosome instead of two. Both of these situations can result in the birth of an infant with various health issues resulting from their abnormal chromosome number.

Who Could Benefit from CCS?

1. Women of advanced maternal age (40 years of age or more) who are at a much higher risk of chromosomal abnormality due to an aging effect on the egg and the way in which the cells of the embryo divide.
2. Women with unexplained recurrent pregnancy loss or repetitive chromosomally abnormal miscarriages.
3. Repeated failed IVF cycles in spite of replacing what appear to be good quality embryos.
4. Any couple wishing to rule out any risk of a chromosomal abnormality in their embryos or offspring.

How is CCS Performed?

In order to have embryos to test, in vitro fertilization (IVF) must be performed. IVF involves the stimulation of a woman's ovaries with medication. This results in the production of several eggs which are then removed from the ovaries when they reach maturity. Sperm is then added to the eggs in the laboratory, a process known as oocyte insemination. The inseminated eggs are then placed in an incubator in the laboratory and checked the following day to confirm which have undergone fertilization. The fertilized eggs, which are now known as embryos, are then grown for an additional 4 days until they reach the fifth day of embryo development. At this stage, the embryos are known as blastocysts.

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NOTE: Not every egg removed from the ovaries will undergo fertilization and not every egg that fertilizes will develop into a blastocyst. It is for this reason that several eggs are required for a successful CCS cycle.

Several cells are then removed from the outer portion of the blastocyst by a process known as "embryo biopsy". These cells represent a sample of those cells that will subsequently develop into the placenta (these are called trophectoderm cells) and have the same genetic makeup as the fetus. Based upon our current knowledge and clinical experience, embryo biopsy does not damage the embryo nor does it have any negative effect on the baby. Once the embryos are biopsied they are immediately frozen by a process known as "vitrification" which is the newest development in embryo freezing and results in a very high survival rate at thawing (approximately 95% or more). In other words, embryos are rarely damaged by the freezing process.

The cells that are removed from each embryo are sent for diagnosis to a specialized genetics laboratory in the United States. Once the results are obtained from the laboratory, we then know which embryos have the normal amount of chromosome material and which are abnormal. The abnormal embryos are not utilized and are discarded. The normal embryo(s) can be replaced in one or more subsequent menstrual cycles.

NOTE: Embryos are biopsied and then frozen while the biopsied cells are analyzed for their chromosome status.

There is an additional advantage to replacing cryopreserved or frozen embryos as opposed to replacing embryos in a fresh IVF cycle. In a frozen embryo replacement cycle the hormonal environment in the woman's body is in a much more normal state than in a fresh cycle where the hormone levels are very high. It has been suggested that replacing embryos in such a more natural environment actually results in better chance of pregnancy.

NOTE: Replacement of thawed embryos which have been vitrified should result in as high a pregnancy rate as those replaced in a fresh cycle.

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Damage to an embryo is extremely rare during the process of embryo biopsy.

1. **T**here is a possibility that no genetically normal embryos will be available for replacement into the uterus following PGS testing.
2. There is a possibility that none of the eggs obtained from the IVF procedure will reach the blastocyst stage. In this situation the biopsy cannot be performed and no embryos will be available for replacement into the uterus.
3. Occasionally, the test does not give us a definite answer regarding the amount of chromosomal material that is in the embryo (1-2% of embryos tested).
4. A frozen embryo, which is genetically normal, may not survive the freeze- thaw process and will thus, be lost from the pool of normal embryos (3-5% risk).
5. There is a 1-2% risk of misdiagnosis. For that reason, prenatal testing is still recommended for any pregnancy following PGS.

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