Chromosome and Genetic Screening

Please spend some time thinking about the options available to reduce the risk of a genetic problem in your child. We will have an opportunity to discuss this in your consultations as you go through treatment.

Screening for chromosome anomalies in patients undergoing IVF

Chromosomes are groups of thousands of genes that contain all the information required to make proteins. We all have 46 chromosomes plus sex chromosomes ie., a female is 46XX and a male is 46XY. I screen all patients for chromosome anomalies at the beginning of the fertility journey. This is called a **Karyotype** and is covered by Medicare. Approximately 1% of patients will have n abnormal karyotype.

Even though couples have normal chromosomes there still is a chance of your embryos being affected by a spontaneous chromosome anomaly that arise during the division process (meiosis) when eggs are produced. Abnormal chromosomes in the embryo can cause failure of treatment, miscarriage or a baby being born with a chromosome problem such as Down's syndrome.

Most of my patients (95+%) opt to screen their embryos during the IVF cycle for spontaneous chromosome anomalies that arise from the egg. Approximately 20% of embryos will have chromosome anomalies in women in their 20's. This rises to greater than 50% in women in their 40's.

Screening embryos for chromosome problems is called preimplantation genetic screening (PGS). PGS improves the chance of having a successful outcome per embryo transferred. It minimises the risk of miscarriage. It also reduces the low risk of a child being born affected by a chromosome anomaly such as Down's Syndrome.

There is an additional cost for PGS.

Optional additional screening of couples for single gene defects

In addition to chromosome problems we all have the risk of being carriers of genetic conditions caused by single gene defects. These occur when one out of millions of genes have an error.

To be affected a child needs to inherit two copies of the abnormal gene., ie inherit one abnormal gene for the same condition from each parent. These are called autosomal recessive conditions. This is an option for screening couples to see whether they are carriers of recessive conditions.

The most common recessive condition in white Australian couples is **cystic fibrosis**. It is possible to screen the female partner for approximately $350. If one partner is found to be a carrier we can then screen the other partner to see whether the baby is at risk.

A more detailed test for a large number of recessive condition is available. This screens for over 100 different recessive conditions including cystic fibrosis. If you decide to go down this more detailed options it is possible to screen the female partner for $579 (approximately due to exchange rate with US$). If one of you is found to carry a particular gene we can then screen the other partner to see whether the baby is at risk. **This test is called Counsyl Family Prep Screens**.

Whether to test the female first or both have a test?

Screening the female partner first and then escalating to screening the male partner is a gene defect is detected to the most cost-effective approach. There will however be a delay in starting your treatment each tests take three weeks.

Some couples opt to both get tested to minimise the delay. The cost higher to do two tests. This is a personal choice.