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fertility matters @ wesley
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Genetic Screening & Diagnosis

- Dr Michael Gattas, Clinical Geneticist, Brisbane Genetics

Pre-Implantation Genetic Screening and Pre-Implantation Genetic Diagnosis

Improvement in embryo culture techniques has meant that embryo, or polar body biopsy, can be used as a source for chromosome or DNA for analysis. This technology has been used in several applications. First to try and improve the success rate for IVF by selection of healthy embryos without any obvious chromosome defects. This is called preimplantation embryo screening (PGS). This patient group tend to be older and have not had success with conventional IVF in the past.

Secondly the technology has been used to avoid the transmission of single gene (autosomal or X linked) disorders in carriers or individuals affected with inherited conditions. This is called preimplantation genetic diagnosis (PGD). Thirdly the technology has been used to select compatible sibling donors for a child who is affected with a life threatening disease that is potentially curable with haemopoietic stem cell transplantation. These disorders are often Mendelian in inheritance (for example thalassaemia or Fanconi anaemia) and HLA compatibility selection is one component of PGD to avoid the birth of another affected child. This tends to take place in a younger patient population with the advantage of proven fertility.

The technology has been controversial as it can be used for social sex selection or reasons of "gender balancing" of offspring. This use is currently not available in Australia because of a voluntary code of practice as recommended by the current NHMRC Guidelines on the Ethical use of Assisted Reproductive Technology. The use of this technology for sex selection is available overseas.

Early Experience

Nearly 20 years since PGD was first used successfully, multicolour FISH analysis for chromosome aneuploidy for chromosomes 13, 18, 21, X, and Y was first used in clinical practice. This expanded to include chromosomes 16 and 22 as abnormalities in these chromosomes are common cause of early miscarriage.

FISH probes for all chromosome telomeres are available and this allows the application of PGD to couples with balanced chromosome translocations who are prone to increased rates of miscarriage.

It soon became apparent that a high proportion of human embryos have aneuploidy at the earliest stages of blastocyst development. This proportion in a healthy couple is probably around 30%, and in an infertile or older couple can increase 2 or 3 fold. There is also frequent chromosome mosaicism at the very earliest stages of human development. This means that two or more cells biopsied from the same embryo may have discordant results. This does make interpretation of PGD results difficult and also creates the potential for both false positive and false negative results for chromosome aneuploidy in a foetus.

Current Practice

The inherent nature of PGD and PGS is that it involves a very small sample for analysis. It is at best around 98% accurate and for this reason clinically important results should be checked by CVS at a 12 week stage of pregnancy. The gold standard for checking chromosome results in an unborn child remains either chorionic villus sampling (CVS) or amniocentesis.

PGD does have application for monogenic disorders where the precise gene fault is known and parental samples are available for linkage analysis. Technically it is demanding and some IVF Units only offer PGD for a limited range of common conditions, such as cystic fibrosis or Huntington disease.

PGS has not been shown to improve success rates in large randomised studies. Supporters of PGS argue that in selected patients it still has a role. The use of newer genome testing methods for PGS may have some utility in this area. Other methods of assessing embryo viability such as simpler biochemical measurements of embryo metabolism continue to be explored.

Problems in Practice

Both PGD and PGS add several thousand dollars to the cost of IVF. This therefore limits their availability to patients with the financial means to pay for these tests. Funding for randomised clinical trials is also lacking, and expertise in molecular genetics analysis and genetic counselling in this area is also in short supply.

Twins or other multiple pregnancy remains the main risk from PGD and other assisted reproduction intervention. Twin pregnancy increases the risk of morbidity to the foetus, neonate, and mother. Twinning, intracytoplasmic sperm injection, and/or in vitro culture through to a 5 day old blastocyst also seem to increase the risk of rare disorders of imprinting such as Beckwith Wiedemann syndrome.

Conclusion

The implementation of a successful PGD and PGS program requires a high level of communication between IVF Specialist, Genetic Specialist, IVF Scientist or Embryologist, and Molecular Genetics laboratory. Most of this dialogue takes place several months before any stimulated cycle can commence.

A patient who maybe interested in using this option is best referred to the Wesley Monash IVF Service as early as possible. Success in achieving a pregnancy with IVF and embryo selection becomes increasingly difficult for woman after the age of 37.

IVF unit welcomes female Clinician

Wesley Monash IVF welcomes another female Clinician to the IVF team!

Dr George is an experienced Clinician from Vellore in India where she worked as Professor of Obstetrics and Gynaecology in the Christian Medical College.

After recently moving to Brisbane with her husband, Dr George has joined the team at Wesley Monash IVF, located at the Wesley Hospital, to offer a fertility and gynaecology service. Dr George is a graduate of the Jawaharlal Nehru Medical College and completed her training in Obstetrics and Gynaecology at the Christian Medical College, Vellore, India in 1989.

As part of her overseas training to obtain her FRANZCOG qualification, Dr George worked in the Reproductive Medicine Unit of Adelaide's Queen Elizabeth Hospital as a Research Fellow and RMO and at the Modbury Public Hospital Adelaide as a Registrar.

Her main interests are in fertility medicine encompassing reproductive endocrinology and IVF treatment cycles and she has a special interest in Male Infertility and Polycystic Ovarian Syndrome.

Dr George is experienced in minimally invasive surgery as it relates to women's general health and fertility and in particular the laparoscopic treatment of endometriosis.

Dr George MBBS, DGO, MD, FRANZCOG will consult from rooms located at the Wesley Hospital currently shared by Drs Allan, Chenoweth and Baines and is happy to take consultations for gynaecology and infertility.

For appointments:
Dr Susan George
(07) 3232 7090

Wesley Monash IVF Clinicians

Wesley Monash IVF is proud to have a dedicated team of highly skilled and experienced Fertility Specialists. All have specialist obstetrician and gynaecological qualifications and have developed their special interests in infertility areas over many years.

Dr John Allan
Medical Director
T: 07 3232 7090
Wesley Campus

Dr John Chenoweth
Deputy Medical Director
T: 07 3232 7626
Wesley Campus

Dr Stephen Baines
T: 07 3232 7144
Wesley Campus, Maryborough

Dr Stephen Cook
T: 07 3371 1777
Wesley Campus

Dr Ross Turner
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Dr Dana Moisuc
T: 07 5478 3533
Buderim

Experience the Wesley Monash IVF Difference

- Experience & Expertise
- Responsible & Ethical Management
- Personalised, Caring Service
- Central Location & Facilities
- Counselling & Support
- Success with more than 2000 babies

Dr Susan George
T: 07 3232 7090
Wesley Campus

Our Doctors

Dr John Allan
Dr John Chenoweth
Dr Stephen Baines
Dr Stephen Cook
Dr Susan George
Dr Dana Moisuc
Dr Ross Turner

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