

# PATHOLOGY FOCUS

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Antenatal Edition | Medical Newsletter 11

## Featured articles:

- Female Reproductive Hormones and Fertility Testing
- Fasting Blood Tests for Lipids? Not Anymore
- Pre-Conception Testing as Preventative Medicine
- State Update

## COVID-19 Update

This year, the restrictions put in place to stop the spread of COVID-19 quickly forced us to change the way healthcare services are provided in Australia.

We saw a dramatic increase in the number of our referrers opting to use telehealth consults, via phone or video, for patient appointments, with the previous face-to-face method of consults only being offered where necessary. So, we knew we had to adapt quickly to support you in ensuring your patients could still access pathology services.

An electronic pathology request form was added to our website and a Telehealth button made available on eOrders (for MedicalDirector Clinical 3.18+), allowing pathology request forms to be emailed directly to patients.

We also wanted to ease patients' concerns of visiting collection centres during COVID-19 and so we implemented strict cleaning protocols and social distancing measures in all our collection centres. When we began collecting pathology specimens for COVID-19, in selected collection centres, strict PPE and patient collection procedures were put in place.

As Australia's response to the COVID-19 pandemic continues to change, our focus will remain on providing you with the best pathology services and diagnostics available, to help you manage the health of your patients. We thank you for continuing to support Clinical Labs throughout this challenging time.

*Article continued on back page.*

# Female Reproductive Hormones and Fertility Testing

By Associate Professor Mirette Saad



## Reproductive Hormones

Reproductive endocrinology encompasses the hormones of the hypothalamic-pituitary-gonadal axis and the adrenal glands. These hormones are crucial for proper reproductive function and include Gonadotropin-Releasing Hormone (GnRH), Lutenising Hormone (LH), Follicle Stimulating Hormone (FSH) and a multitude of sex steroids.

LH primarily stimulates the production of hormones by the gonads whereas FSH stimulates the development of the germ cells. The sex steroids are synthesised at the ovaries, testes, and adrenal glands and are responsible for the manifestation of primary and secondary sexual characteristics. The thyroid gland produces hormones that are essential for normal metabolism, hormone production and fertility. This review article discusses female reproductive hormones and their role in fertility testing.

## Physiology of Ovarian Cycle

Normal women display considerable variation in cycle length, which varies from 25-30 days, with a normal ovulatory menstruation cycle of 28 days, on average.

Oestrogens, progesterone and androgens are secreted by the ovarian follicles of the ovaries. With a normal ovarian reserve, oestradiol (E2) produced by the ovarian follicles and the corpus luteum increases, reaching a mid-cycle peak then fall off abruptly after ovulation. This exerts negative feedback on the hypothalamic-pituitary axis to stabilise the level of FSH and LH release, such that one dominant follicle is selected each cycle. A mid-cycle surge in LH production stimulates a series of events that culminate in ovulation, with FSH levels falling after this event.

*"Normal women display considerable variation in cycle length, which varies from 25-30 days, with a normal ovulatory menstruation cycle of 28 days, on average."*

Any injury to the hypothalamus, or the presence of either psychological or physical stressors, lead to changes in these hormonal cues, resulting in anovulation and amenorrhea.



## Ovarian Reserve and Anti-Müllerian Hormone (AMH)

In females, Anti-Müllerian Hormone (AMH) is an established biomarker, produced by antral and pre-antral follicles for assessing ovarian reserve, which is considered an important tool in assessing potential fertility. AMH can help in the assessment of reproductive capacity and in the prediction of oocyte retrieval (egg collection) and ovarian responsiveness to stimulation regimes.

*"AMH can help in the assessment of reproductive capacity. Low serum levels of AMH is suggestive of poor ovarian reserve, whereas high levels may indicate a polycystic ovary syndrome (PCOS)."*

## Menopause and FSH Hormone

Menopause is a normal change that women go through when the ovarian reserve has been depleted. Normally, menopause occurs within a 10-year window, from 45-55 years of age, with an average age in Australia of 51.5 yrs.

**Fluctuating levels of FSH and oestradiol** can be noticed, during both peri-menopausal and pre-menopausal stages, hence, they are not considered reliable predictors of menopause, as they are sometimes at pre-menopausal levels.

## Infertility or Subfertility: Definition and Subtypes

One in six Australian couples experience trouble conceiving. Infertility is defined as a unique medical condition because it involves a couple, rather than a single individual. WHO defines infertility as failure of a couple to conceive after 12 months of regular intercourse without use of contraception in women less than 35 years of age; and after six months of regular intercourse without use of contraception in women 35 years and older.

**Primary infertility** is infertility in a couple who have never had a child. **Secondary infertility** is failure to conceive following a previous pregnancy and is considered the most common form of female infertility, mainly due to reproductive tract infections. It is estimated that male infertility represents 20-30% of cases, while 20-35% of cases are due to female infertility and the rest are due to combined (~20%) or unexplained reasons (~15%).

## Fertility Clinical Assessment in Couples

For conception to occur, viable spermatozoa, ovulation and functional anatomy are required. In cases of infertility, both partners should be investigated.

A couple who is unsuccessful at achieving pregnancy should seek a comprehensive physical and medical assessment by clinicians in order to identify causes that

can be medically intervened. In addition to structural and surgical causes, laboratory investigations are essential to identify clues to the underlying aetiology to reach a clinical diagnosis.



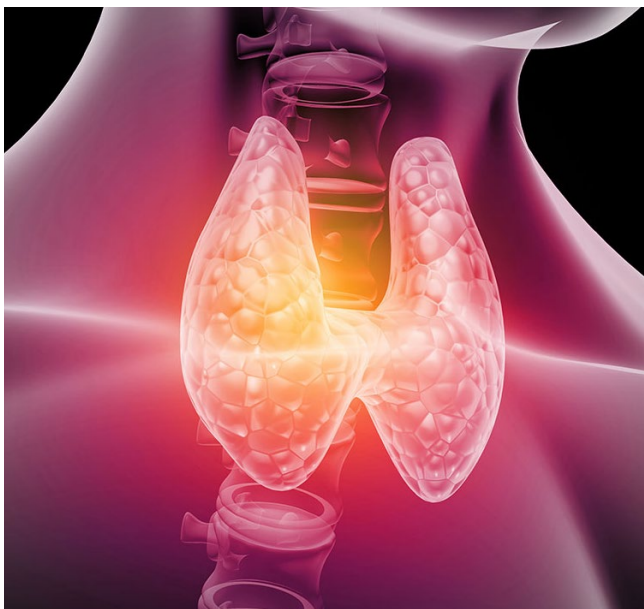
### Investigations for Male Infertility

- Semen analysis is an essential component of fertility with an utmost predictive value
  - repeat after 6 weeks, if abnormal
- LH, FSH, testosterone
- Prolactin, thyroid function
- If azoospermia present
  - cystic fibrosis screen, karyotyping (for conditions such as Klinefelter's syndrome (47, XXY) and others
  - testicular biopsy

*“A couple who is unsuccessful at achieving pregnancy should seek a comprehensive medical assessment. Laboratory investigations are essential to identify clues to the underlying aetiology to reach a clinical diagnosis.”*

### Investigations in Female Infertility (Table 1)

Investigations for anovulation include: FSH, LH, Prolactin, TSH, Testosterone, SHBG, Trans-vaginal U/S, CT/MRI pituitary, Karyotyping.



### Pituitary and Ovulatory Biomarkers:

Common causes of anovulation include polycystic ovary syndrome (PCOS), hypothalamic amenorrhoea (HA) and premature ovarian insufficiency (POI).

- **Ovulatory Function:** FSH/LH and Oestradiol (E2) are essential in assessing the ovulatory cycle. They are best evaluated in the early follicular phase (day 2-6). An elevated FSH level (>10mIU/mL) early in the follicular phase is indicative of a low ovarian reserve.
- **Ovulation Predictor:** evaluating the LH surge is the best single assay, while the measurement of LH plus pre-ovulatory oestrogen is the best prediction.
- **Prolactin Testing** is required in order to exclude hyperprolactinaemia. It can be measured anytime during the day or in the menstrual cycle (but not after exercise or stress). Consider a follow up CT/MRI of pituitary if pituitary macroadenoma is suspected, particularly in the context of amenorrhoea, galactorrhoea, visual field abnormality or headaches, which warrant referral to endocrinology.
- **TSH** is also recommended to exclude hypothyroidism.

### Ovarian Reserve and AMH Testing:

Low serum levels of AMH is suggestive of poor ovarian reserve, whereas high levels may indicate a polycystic ovary syndrome (PCOS).

### Primary Amenorrhea:

- After excluding pregnancy, pituitary and ovarian hormones' testing is warranted (see above).
- Karyotyping chromosomal/genetic analysis should be performed in women with ambiguous genitalia or evidence of primary ovarian insufficiency (hypergonadotropic hypogonadism) or clinical features suggestive of Turner syndrome (45,X0).

### Secondary Amenorrhea:

- **Pregnancy:** Human chorionic gonadotrophin (HCG) where a result >25 IU/L (normal <5) is indicative of early pregnancy.
- **Menopause:** where monitoring check is required for both FSH and E2.
- Other medical causes should be investigated.

### Androgen Testing in Hirsutism:

While hirsutism in females is most commonly idiopathic in aetiology (60% of cases), 40% of cases can be due to elevated androgen levels. Female androgens are produced the ovaries and adrenal glands.

True androgen excess (including total plasma testosterone, DHEA, 17-OH progesterone (17-OHP) and androstendione), can occur in ovarian and adrenal gland disorders. High androgens can be found in PCOS in about 35% of cases, while 5% of cases are due to other metabolic conditions, such as congenital adrenal hyperplasia (CAH), Cushing syndrome, hyperprolactinaemia, and adrenal and ovarian tumours.



## Polycystic Ovary Syndrome (PCOS):

- In PCOS cases, in addition to the above hormonal testing, ovarian ultrasound is usually recommended.
- A follow-up testing for early diagnosis of diabetes mellitus and hyperlipidaemia, including fasting glucose, insulin, HBA1c, oral glucose tolerance tests (OGTT), insulin resistance and lipid levels, is recommended.

Blood Test	When	Results' Interpretation
Progesterone	D21 in a regular cycle or 7 days prior to expected menses if cycle not 28 days	>30 nmol/L confirms ovulation, whereas low values indicate anovulation and a need for follow-up testing
FSH	Day 2-6 (Early Follicular Phase, best on Day 3)	Raised serum FSH (>10 IU/L) during the early follicular phase is reflective of reduced ovarian reserve, albeit it is a relatively late feature
LH	Early Follicular Phase (Day 2-6)	>10 may be seen in PCOS
Prolactin	Anytime of the cycle	>1000 (mIU/L) may indicate pituitary macroadenoma
AMH	Anytime of the cycle; a sensitive hormone marker of fertility potential	Low values may indicate poor ovarian reserve while high values may suggest PCOS
Testosterone	Anytime of the cycle	May suggest PCOS or CAH (17- OHP and DHEA level should be assessed during the follicular phase)

Table 1: Summary of Some Fertility Hormones Testing in Women.

## General Workup Investigations

General causes of infertility should be investigated, such as anorexia nervosa, malnutrition, substance abuse, alcoholism, exogenous steroids, tumours/chemotherapy, infections and metabolic conditions (including diabetes mellitus and coeliac disease), and chronic illness such as HIV, hepatitis, sexually transmitted diseases (STDs) and sarcoidosis.

## Blood Investigations and Follow-up Management

### May Include

- General chemistry, including liver function and kidney function tests
- Nutritional Assessment: Vitamins, iron studies and trace elements
- Tumour markers
- Angiotensin-converting enzyme
- Metabolic/endocrine screening: cortisol and IGF-1, haemochromatosis, coeliac autoantibodies and genetic testing
- Drug/alcohol screen
- Chronic infections and STD screen
- Imaging workup in hyperprolactinaemia, PCOS, adrenal and ovarian tumours
- Referral to an endocrinology specialist service for further assessment, if indicated

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# Fasting blood tests for lipids? Not anymore.

By Dr David Deam

Recent evidence is challenging the need to have patients fast for blood tests when assessing for risk of cardiovascular disease.

It has now been shown that non-fasting triglycerides are superior to fasting in predicting cardiovascular risk. Patients usually eat regularly during the day with the fasting state only occurring shortly before breakfast; a lipid profile measured during this time may not accurately reflect the lipid concentration during the day. In addition, lipids and lipoproteins only change minimally in response to normal food intake; for instance, in four large prospective studies, there was minimal change in triglyceride levels in patients who had a non-fasting versus a fasting blood test.<sup>(1, 2)</sup> Further, in results from a population of 108,602, patients who had non-fasting random blood tests and had the highest lipid results (including triglycerides) had the highest risk of ischaemic heart disease (IHD).<sup>(3)</sup> Finally, results from studies assessing the effectiveness of lipid-lowering agents, using non-fasting samples, show that reducing the levels of non-fasting lipids reduced the risk of cardiovascular disease.<sup>(4)</sup>

There is no sound scientific evidence as to why fasting should be superior to non-fasting when evaluating a lipid profile for cardiovascular risk prediction. Non-fasting tests simplify blood tests in the laboratory, avoid the inconvenience of fasting tests and prevent the clinical risk associated with fasting blood tests in patients with diabetes who may be at risk of iatrogenic hypoglycaemia. The scientific evidence suggests that we should move away from fasting blood tests, despite the fact that this is the way it has always been done; fortunately, guidelines are being modified to reflect these changes.<sup>(5, 6)</sup>

## In Summary

1. Lipids only differ slightly in fasting versus non-fasting tests
2. Non-fasting triglycerides are better at predicting cardiovascular risk
3. Studies of lipid-lowering agents using non-fasting tests showed reducing levels of non-fasting lipids correlated with reduced risk of cardiovascular disease
4. Non-fasting lipids are more convenient and safer for patients (particularly for diabetics)
5. Guidelines are currently being updated to reflect the above

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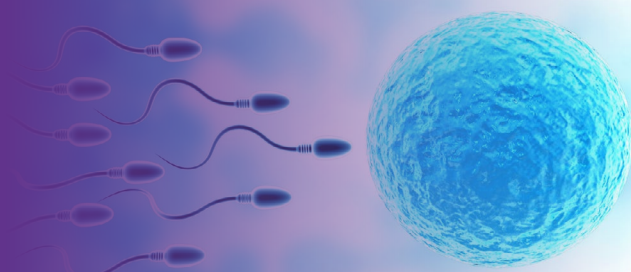
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Dr Deam graduated with Honours in Medicine from Monash University in 1978 and obtained his FRCPA in 1985, following postgraduate training in Biochemistry at the Royal Melbourne Hospital. After several posts in Chemical Pathology at the Royal Melbourne Hospital and the Royal Women's Hospital, he was appointed Head of Chemical Pathology at the Royal Melbourne in 1996. He joined Gribbles Pathology (now Australian Clinical Labs) in 1998. Dr Deam has played an active role in teaching scientific, nursing and medical staff at both undergraduate and postgraduate levels and has been an examiner for the Australasian Association of Clinical Biochemists as well as the Royal College of Pathologists of Australasia. Dr Deam's research interests and publications include work on thyroid function testing, various aspects of diagnostic protein measurement and the rational use of biochemical tests.

# Pre-Conception Testing as Preventative Medicine

By Dr Lionel Wu



Dr Lionel Wu is a general practitioner and principal of Medifirst Family Clinic in Melbourne's south-east. Dr Wu obtained his Diploma in Paediatric Qualifications in 2011 through Sydney University's Westmead Hospital. He has a special interest in paediatrics and obstetrics and has attended many courses and seminars in these specialist areas. Dr Wu opened MediFirst Family Clinic five years ago to cater for the multicultural demographic in the clinic's surrounding area and to aid patients in proactively managing their health.

In this article, Dr Wu discusses why pre-conception testing, including fertility testing, is essential in managing a patient's physical and mental health.

**Pre-conception testing for women is vital, as it acts as a measure by identifying potential health issues which can be treated or resolved early, thus preventing pregnancy-related issues that may arise in the future.**

At Medifirst Family Clinic, we see a lot of women for pre-conception planning and pregnancy management. About 30% of our patients are first-time mums, or the second- or third-time mums of our paediatric patients. The women that come for a pre-conception planning appointment often ask me about what tests they require and what they need to do in order to get pregnant.

Below, I outline and discuss the tests I always offer to patients considering a pregnancy, including some nonroutine tests that I believe are equally as important.

## Grouping pre-conception tests into three important categories

I have divided the following pre-conception pathology tests that I routinely order for my patients into three groups.

Group 1 - General wellness tests	FBE (full blood examination), UE (kidney function), LFT (liver function test), haemoglobin electrophoresis, B12, iron studies, TFT (thyroid function test), Vitamin D, blood group and antibodies, FSH (follicle-stimulating hormone), LH (luteinizing hormone), 21st day progesterone level, oestrogen, pap smear (if not up to date).
Group 2 - Screening tests for blood antibodies and infectious diseases	Measles, mumps, rubella and varicella serology. Parvovirus antibodies, CMV (cytomegalovirus), EBV (Epstein-Barr virus) serology, HIV, hepatitis ABC serology, syphilis and toxoplasma serology. STIs (chlamydia and gonorrhoea), urine MSU MCS.
Groups 1 & 2 are all ordered at the first pre-conception appointment.	
Group 3 - Specific fertility tests	Anti-Müllerian hormone (AMH), pelvic ultrasound (fibroid, endometriosis, adenomyosis, polyps). If I suspect patient of having polycystic ovarian syndrome (PCOS) - fasting LDL, HDL, TG, BSL, insulin, HbA1c; LH:FSH, androgen studies, SBGH (sex hormone binding globulin), bHCG, TSH (thyroid stimulating hormone), cortisol, prolactin, 17-hydroxyprogesterone, DHEAS

## Group 1: First things first, testing for overall wellness

### Thyroid function

I always check a patient's thyroid function as part of the pre-conception general wellness tests because thyroid hormone imbalances can affect a woman's fertility. If a woman doesn't have enough thyroid hormones in her body, this will affect her ovulation and fertility. In a male, low levels will also affect sperm production. Having healthy thyroid hormone levels in both parents is extremely important for a successful conception. An underactive thyroid can be treated with hormone replacement tablets, however, the patient will require continual monitoring throughout her pregnancy while undergoing treatment.

### Sex hormones and their roles

As part of pre-conception screening, I also include tests to measure important hormones.

**Follicle-stimulating hormone (FSH)** is the first in a cascade of hormones that are necessary to launch a patient's pregnancy, and FSH is present before she even conceives.

**Luteinizing hormone (LH)** works in concert with FSH to orchestrate the menstrual cycle. Both FSH and LH are inactive during pregnancy, itself.

**Progesterone** is a hormone produced by the ovaries and the placenta during pregnancy. It stimulates the thickening of the uterine lining for implantation of a fertilized egg. Normally, a 21st day progesterone level is ordered to check if a woman has successfully ovulated.

The **oestrogen group of hormones** helps develop female sexual traits. It is normally formed in the ovaries. It is also made by the placenta during pregnancy to help maintain a healthy pregnancy.

### Folate and fetal spinal cord development

One of the first things I check is whether the patient has adequate levels of folate in her body. Insufficient folate levels can result in developmental problems, such as spina bifida. The current recommendation is for folate to be taken three months before conception; so, the minute a woman comes in for her pre-conception appointment, I will recommend that she starts taking folate immediately.

### Iron deficiency resulting in fatigue and anaemia

Ensuring a woman has adequate iron levels before pregnancy is very important, as iron deficiency is a commonly seen problem in pregnant women – particularly towards the later stages of pregnancy. Pregnant women

can become severely iron deficient, leading to anaemia. This can result in them becoming very tired and fatigued and, in certain cases, even experiencing trouble with breathing.

This is why I routinely monitor a woman's iron levels at different stages of her pregnancy. If the patient is iron deficient, I generally recommend that she increases her meat intake as well as take an iron supplement continuously, until she gives birth.

*'As general practitioners, I think we are like the gatekeepers of the community's health – the go-to persons with regard to their health issues.'*

### Timely cervical screening test

As part of the pre-conception general wellness tests, I always recommend a cervical screening test with STI (sexually transmitted infection) screening, as both can be performed at the same time. We have two female doctors at Medifirst Family Clinic who can perform this test, if patients are comfortable with having it done. Generally, most obstetricians would like their patients to be up-to-date with their Pap smears before they fall pregnant. As general practitioners, we want our patients to take all the recommended routine tests to ensure that they are managing their ongoing health and as a preventative measure for their general well-being.

### Screening for hereditary blood disorders

In addition to the routine general wellness tests, such as FBE and those listed above, another test I always recommend is haemoglobinopathy screening, as some patients may be carriers of conditions, such as thalassemia or sickle cell anaemia, without being aware of it. If both parents are carriers of thalassemia, there is a one-in-four risk that their baby may be born with the disease, which can have devastating and lethal consequences. Thalassemia is more common in Asian, African and Mediterranean cultures, which is why I offer this test to all patients, given Melbourne's very diverse population. This testing is separate from genetic carrier screening, which I also offer my patients.

## Group 2: Screening for infectious diseases

### Devastating effect of infections on pregnancy

During their pregnancy, it is essential that women are protected from infections, such as **measles, mumps, rubella, and varicella**. These diseases have devastating effects by causing life-long disabilities on unborn babies, such as malformed limbs or permanent deafness or blindness. Therefore, it is vital for pregnant women to get immunised if they don't have enough protection to carry them through their pregnancies.

The same goes for **hepatitis A and B**. I would strongly encourage women to protect themselves through immunisation.

When ordering tests to screen for infectious diseases, I also routinely run an immunity check for **parvovirus**, which is commonly known as 'slapped cheek' syndrome. If a patient contracts this infection while she is pregnant, the likelihood of a miscarriage increases dramatically, making this a highly important pre-conception test to have.

**Toxoplasmosis** is another infection that increases the risk of miscarriage in pregnancy. It is caused by a common parasite, toxoplasma, that resides in the gut of cats. In general, if you contracted toxoplasmosis before becoming pregnant, your immunity will protect your unborn child. Some experts suggest a 6-month wait after a recent infection before attempting to conceive. I normally recommend that if a woman has a cat at home, her partner should undertake the feeding and cleaning of the pet.

**Screening for STIs** (sexually transmitted infections), in particular **chlamydia and gonorrhoea**, is also very important, as these infections may cause PID (pelvic inflammatory disease). It is a major cause of tubal pregnancy (ectopic pregnancy). An ectopic pregnancy can occur when untreated PID causes scar tissue to develop in the fallopian tubes. The scar tissue prevents the fertilized egg from making its way through the fallopian tube to implant in the uterus. Scarring of the fallopian tubes may also cause subfertility in some women.

### Rescreening high-risk women for syphilis in the 3rd trimester

There has been an alarming increase in the number of cases of infectious syphilis reported in Victoria, with numbers nearly tripling from 634 in 2014 to 1670 in 2019. Of note is the number of women of reproductive age, in their 20's and 30's, contracting the infectious disease. If a woman contracts syphilis while pregnant and the disease is left untreated, the effects can be catastrophic. Congenital syphilis can lead to miscarriage, stillbirth, premature birth, low birth weight and other health issues in the baby. In July this year, Victoria's Chief Health Officer, Professor Brett Sutton, issued health professionals with new advice for testing pregnant women for syphilis.

#### Latest antenatal testing advice for syphilis:

- Screen all pregnant women for syphilis during routine antenatal testing in the first trimester
- Re-screen women at high risk of contracting syphilis again between 28-32 weeks and at delivery
- Re-screen women in later pregnancy with clinical signs of any sexually transmitted infection, including a discharge or ulcers

#### Treatment and follow-up

To avoid congenital syphilis, the disease should be treated with long-acting (benzathine) penicillin. Partner tracing and treatment is also required. Babies born to mothers with syphilis in pregnancy should be followed up by a specialist.



NEW

### Group 3: Specific fertility tests

#### Why early fertility testing is so vital

Many fertility issues can be dealt with, treated or even cured, if they are diagnosed early. If not, the financial as well as the psychological and emotional devastation on the patients can be phenomenal. The impact on their well-being is similar to that of someone who has had a heart attack or stroke. This is why it is so important to identify potential problems as early as possible.

When we check and monitor patients' cholesterol levels, we hope to prevent heart attacks or strokes. Similarly, when we conduct early tests and screening for fertility issues in women, it is to avoid a lot of heartache later on.

#### Patient age and fertility

If a patient has difficulty conceiving, I will advise a fertility test. I will take into account the patient's age and how long she has been trying to conceive.

#### Patients under the age of 30:

Will be offered fertility testing if they have been actively trying for a year without success.

#### Patients over the age of 30:

Will be offered fertility testing if they have been trying for six months unsuccessfully.

#### Recommended fertility tests

These are the fertility tests that I order for my female patients:

- Anti-Müllerian hormone (AMH), to check the woman's ovarian reserve
- Pelvic ultrasound (US), to screen for endometriosis, adenomyosis, fibroid and polycystic ovarian syndrome (PCOS).

#### Polycystic ovarian syndrome – fertility issues explained

In addition to routine fertility hormone tests, I also run androgen tests to check for PCOS which is caused by an imbalance of reproductive hormones. The hormonal imbalances can interfere with the growth and release of eggs from the ovaries and be a potential cause of infertility.

#### The importance of pre-conception testing

As general practitioners, I think we act like the gatekeepers of the community's health – we are their go-to persons in regard to their health issues.

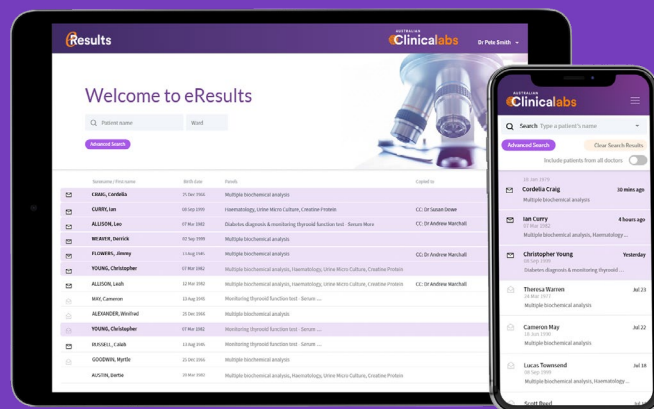
We perform preventative medicine, such as tests for cholesterol and diabetes, to help prevent heart attacks and strokes. At the same time, and equally important, we help women and couples who want to have a family but have trouble conceiving.

And, by advising them on fundamental tests that are needed early, we fulfil a responsible role in helping them prepare for a healthy pregnancy and baby.

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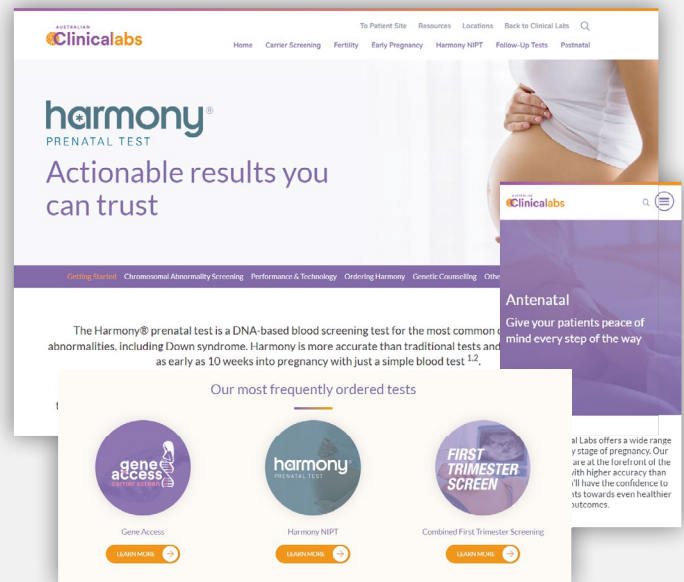
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# Genetic Carrier Status and Early Pregnancy Screening

by Dr Stuart Prosser

At One for Women, we are increasingly seeing patients early in their pregnancy - prior to 10 weeks' gestation. This is fantastic as it allows us to undertake early pregnancy screening and education.

The initial consultation involves assessment for pre-eclampsia risk, cervical length shortening risk and importantly involves a discussion around genetic carrier screening and first trimester screening. While most patients are aware of first trimester screening with a large move towards NIPT and structural ultrasound over the last 3 years, the understanding and knowledge of genetic carrier screening remains low despite a RANZCOG recommendation that all pregnant women should be offered this test.

Ideally, we would like to discuss genetic carrier screening pre-conception to allow the prospective couple to know if they are carriers prior to considering having a baby together. If both partners are found to be carriers of an autosomal recessive condition, or if the female partner is a carrier of an X-linked condition, they have a 1 in 4 chance of each pregnancy being affected by the condition. They are then offered referral to a genetic counsellor to consider their reproductive options.

Once pregnant, options become more limited - diagnostic testing and the difficult decision of whether to continue

with the pregnancy. Each of the genetic conditions causes significant morbidity and leads to shortened life expectancy.

Over the last 12 months, I have seen an increased referral rate for genetic testing. Thankfully, I am starting to capture the patients prior to conception, allowing for screening to be undertaken and appropriate counselling about risk of inherited genetic conditions. In a similar way to NIPT testing, I believe that over the next three years, as the knowledge of the importance of genetic carrier screening increases, the rate of testing will continue to climb.

## About the author



## Dr Stuart Prosser

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Dr Stuart Prosser is the Founder and Medical Director at One for Women and has extensive experience in General Practice, GP Obstetrics and Anaesthetics. He consistently advocates for a multi-disciplinary approach to health care, one that increases the quality of both the patient experience and outcomes.



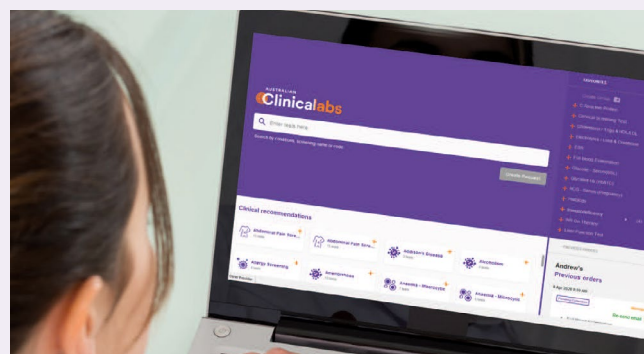
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