

Congratulations on your pregnancy and thank you for choosing Coastal Women's Health for your obstetric care. We value you as our patient and are excited to support you!

Every pregnancy is unique and has its own challenges. We strive to recognise those differences and provide you with outstanding medical care.

Your health and the health of your baby are our priority!

Thank you for allowing our practice to be part of this life changing experience.

Coastal Women's Health

Address: 60 William Street, Gosford

Office hours: Monday-Friday, 9am to 4pm

Phone: (02) 4324 1992

During non-office hours, the above number will automatically reach the answering service, and you will be prompted to leave a message. Please speak slowly and clearly when leaving your name and phone number.

If you have an emergency, please go to your hospital (Gosford Private or Public Hospital) and the midwife will inform Dr Farag.

For all non-urgent questions, we encourage you to email us and send messages through the patient portal.

Dr Farag attends all deliveries at both Gosford Private and Public Hospitals.

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Nutrition and Diet

Balanced nutrition is essential to maintaining good health and is even more important during your pregnancy. The foods that you eat provide the nutrients that will support your baby's growth. In addition, certain stages of fetal growth require higher quantities of certain nutrients. It is important to be mindful of these changes and maintain a healthy diet throughout your pregnancy.

Daily Guidelines

- Breads/Grains/Carbs: 6-11 servings (Make half your grains whole grains!)
- Fruit: 2-4 servings
- Vegetables: 4 or more servings
- Dairy: 4 servings
- Limit fatty, salty, and highly processed foods

Choose whole, minimally processed foods as much as possible. Eat more foods with the following nutrients:

Nutrients	Examples
Fiber	Whole grains and rice, legumes, whole fruits and vegetables
Folic Acid	Dark-green leafy vegetables, legumes, veal, and liver
Iron	Legumes, leafy greens, seafood, meat, eggs, and tofu
Vitamin C	Citrus fruits and cruciferous vegetables

Eating for Two? This is a common myth. Calorie recommendations for pregnant women are only about an extra 300 calories a day on average!

Calorie Recommendations

- About 1,800 calories per day during the first trimester
- About 2,200 calories per day during the second trimester
- About 2,400 calories per day during the third trimester

*If you are pregnant with twins or multiples, you may need to increase your daily calorie intake. Make sure to discuss this with your provider.

Prenatal Vitamins

Prenatal vitamins are supplements that can help you consume the recommended levels of certain vitamins and minerals that can be hard to get through diet alone. The most important nutrients in prenatal vitamins are folic acid, calcium, iron, and vitamin D.

- **Folic Acid:** This is one of the most important supplements during pregnancy. It helps prevent neural tube defects during fetal development. The neural tube becomes the baby's brain and spinal cord and is developed during the first month of pregnancy. It is recommended that you receive at least 500 micrograms (0.5 milligrams) per day. Occasionally you may need extra dose of 5 milligrams e.g., Megafol



- **Calcium:** This important nutrient creates strong bones and teeth for both the mother and the fetus. It is also important for the muscular, circulatory, and nervous systems. It is recommended that you receive 1,000 milligrams per day.
- **Iron:** This nutrient is essential for the development of blood and muscle cells. Iron prevents anaemia (decreased number of red blood cells). It is recommended that you intake 27 milligrams per day. Talk with your provider about whether you need to supplement your iron intake.
- **Vitamin D:** Promotes absorption of calcium in the body. It is recommended that you intake about 400-800 IU per day.

Other nutrients found in prenatal vitamins include DHA/EPA, vitamin C, vitamin E, vitamin B12, Thiamine, Riboflavin, Niacin, and Zinc. Prenatal vitamins are available both over the counter and by prescription. If you experience uncomfortable symptoms like nausea or constipation while taking prenatal vitamins, try taking them at night before bed or switching to a prenatal gummy. Contact your physician for alternative supplement options if your symptoms are intolerable.

Vegetarian/Vegan Diets

There are no significant changes in diet recommendations between a non-vegetarian and a vegetarian diet. However, if you are vegan, you are more likely to have difficulty getting the daily levels of vitamin B12, iron, calcium, and folic acid. Make sure to be mindful of the daily recommendations and find alternative ways to reach these levels, such as daily supplements.

Pregnancy Weight Gain

Weight gain is natural over the course of the pregnancy. However, the amount of weight gained varies from woman to woman. The National Academy of Medicine suggests the following weight gain ranges based on your starting pre-pregnancy body mass index (BMI):

	BMI	Weight Gain
Underweight	< 18.5	12 to 18 kgs
Normal Weight	18.5-24.9	12 to 15 kgs
Overweight	25-29.9	8 to 12 kgs
Obese	> 30	6 to 10 kgs



Weight-Related Complications

More is not always better. Excessive maternal weight gain is associated with an increased risk of pregnancy complications, including diabetes, high blood pressure, and necessity for caesarean delivery. Infants of overweight women are more likely to be bigger and have an increased risk of experiencing birth trauma and not descending into the vaginal canal. This occurs more frequently in women shorter than 160 cm.

Where does the weight go?

- **Baby:** 3.2-3.6 kgs
- **Stores of fat, protein, & other nutrients:** 3.0-3.2 kgs
- **Increased Blood Volume:** 1.8kgs
- **Increased Fluid Volume:** 1.8kgs
- **Breast Tissue:** 1kg
- **Uterus:** 1kg
- **Amniotic Fluid:** 1kg
- **Placenta:** 5 kgs

By following general nutrition guidelines, eating a wide variety of foods, and avoiding junk foods, you stand the best chance of growing a healthy baby.

Foods to Limit/Avoid in Pregnancy

There are certain foods that you should avoid or limit to decrease your risk of illness or complications during your pregnancy. The list below explains in detail what foods to limit or avoid, along with cooking guidelines for your reference. For further information about food risks, visit: [FOOD SAFETY DURING PREGNANCY-THE ROYAL WOMEN'S HOSPITAL](#).

- **Alcohol:** Alcohol quickly passes through the placenta into the baby's bloodstream. When alcohol is consumed during pregnancy, infants may be born with physical, mental, and behavioural problems characteristic of fetal alcohol syndrome. These babies are smaller than unexposed babies and may also have many abnormalities including heart defects and intellectual disabilities.

Since it is unknown how much alcohol puts the fetus at risk for **fetal alcohol syndrome**, it is safest to eliminate alcohol entirely during pregnancy.

- **Caffeine:** There is conflicting and inconsistent evidence that the consumption of caffeine or coffee during pregnancy adversely affects the fetus. It may decrease the availability of certain nutrients such as calcium, zinc, and iron.

We recommend moderate caffeine consumption. One mug of coffee per day appears to be safe. Remember that caffeine is not only in coffee or tea, but it is also found in products such as chocolate, soda, and certain medications. In general, it is best to avoid caffeine consumption during pregnancy, or at the very least limit caffeine intake.

- **Seafood that is high in mercury or uncooked:** Seafood is a great source of protein, omega-3 fatty acids, and iron. We believe women can eat up 2-3 servings of seafood per week safely.



- Fish that are high in mercury should be limited. Mercury interferes with the development of the baby's nervous system.
 - Limit white (albacore) tuna and tuna steaks to 6 ounces (170 grams) a week
 - High mercury-containing fish include **swordfish, shark, king mackerel, marlin, orange roughy, and tilefish.**
 - Cooking: Raw fish and shellfish could contain harmful bacteria/viruses.
 - Only eat seafood that has been thoroughly cooked.
 - Cook seafood to an internal temperature of 60°.
 - Smoked seafood should be cooked to 75°.
- **Undercooked Foods:** These foods pose higher risks of food poisoning and other foodborne illnesses. These guidelines are for everyone, but are particularly pertinent for pregnant women, who are more susceptible to foodborne illness:
 - **Eggs** and foods containing egg products should be cooked to 75°.
 - **Beef, veal, lamb** should be cooked to 65°.
 - **Pork and ground meats** should be cooked to 70°.
 - **Poultry** (and stuffing if included) should be cooked to 75°.
 - **Processed Meats:** There are a lot of opportunities for meat to be contaminated during production, especially if processed. The most common foodborne illness related to processed meats is listeriosis.

Common processed meats include bologna, salami, hot dogs, and other deli meats.

- **Cooking:** Listeria can be killed by heating food to the appropriate temperature. All deli-style meats and poultry (cold cuts, hot dogs, dry sausage, etc.) should be heated to 75° before eating.
- **Unpasteurized Foods:** These foods have higher risks of carrying foodborne illnesses. Examples of unpasteurized foods include:
 - Raw milk (often found in soft cheeses).
 - Brie, feta, camembert, blue cheese, queso blanco, queso fresco.
 - Some fresh/cold pressed juices and ciders.

Read labels on all cheeses, milk products, and juices and do not consume unless pasteurized.

- **Unwashed Food:** Do not consume raw vegetables and fruits without thoroughly washing them first. Make sure to also wash sprouts and other earth plants extensively before consuming them.
- **Large Quantities of Vitamin A:** High amounts of vitamin A can lead to birth defects. Liver is a common food item high in vitamin A and should be eaten in moderation.



Exercise in Pregnancy

Proper exercise increases elasticity and strength of muscles. It reduces your risk of gestational diabetes, pregnancy-related hypertension, and stresses related to pregnancy and labour, and improves your heart and lung efficiency. The American College of Obstetricians and Gynaecologists suggests that women without any medical or prenatal complications engage in regular, moderate to intense physical activity throughout their pregnancy.

Exercise Guidelines

- Discuss pre-pregnancy fitness levels with your care provider to determine an appropriate exercise regimen for your pregnancy.
- An average of 30 minutes a day of moderate aerobic exercise is recommended. Examples include walking, swimming, rowing, stationary cycling, or Pilates.
- Make sure you warm up and cool down before and after your exercise. This is important when exercising during pregnancy because your muscles and joints are more stressed due to metabolic changes occurring in your body.
- Drink plenty of water to avoid dehydration and overheating.
- Wear proper footwear and supportive bras.
- Childbirth preparation exercises are also great to incorporate into your exercise regimen. They will help keep you limber and ease common aches and pains associated with pregnancy.
- Keep in mind that your growing baby will demand more of your energy so you may become short of breath or tired more easily than you were before.

Things to avoid while exercising

- Do not hold your breath during exercises.
- Avoid activities that involve strenuous back work.
- Refrain from participating in sports with high contact, such as ice hockey, basketball, and soccer, as well as activities that have risks of falling such as gymnastics, horseback riding, skiing, and outdoor cycling.
- Do not engage in underwater activities such as scuba diving (the air decompression poses potential risks to the foetus).
- Avoid activities at high altitudes.
- Avoid exercise that raises the core body temperature for long periods of time (i.e., hot yoga).

Exercise warning signs

If you experience any of the following symptoms, STOP your activity and contact your provider immediately:

- Vaginal bleeding/spotting
- Abdominal pain
- Chest pain
- Severe shortness of breath
- Dizziness and/or nausea
- Loss of muscle control
- Contractions
- Severe headaches



Sex in Pregnancy

It's okay to have sex while you're pregnant! However, it's common to have concerns when it comes to engaging in sexual activity during pregnancy. If your pregnancy is proceeding normally and you & your partner feel up to it, you can have sex whenever you like.

Keep in mind that your body is going through changes, such as hormonal fluctuations and fatigue, which could make having sex less desirable. There is considerable variety in sexual interest and expression during pregnancy; some women find it enhanced, while others find that the discomforts of pregnancy diminish their desire for physical intimacy. Many psychological factors also affect women and their partners' interest in sexual expression during pregnancy, including feelings about changes in the woman's body and about becoming parents. Feel free to discuss any questions and concerns about sexual issues with your care provider.

The following paragraphs present a spectrum of feelings a woman might experience regarding sexual arousal and intimacy during the various stages of pregnancy.

- **First trimester:** Increased incidence of significant fatigue and nausea. Sexual interest is generally decreased. Also, there may be a fear of causing a miscarriage. Generally, unless there is a history of cramping or bleeding, there is no contraindication to sexual intercourse and no evidence that intercourse or orgasm causes miscarriage.
- **Second trimester:** As the discomforts of the first trimester resolve, this is often a time of heightened interest in sexual expression. Also, increased blood supply to the pelvic organs due to the changes of pregnancy often makes sexual activity more pleasurable.
- **Third trimester:** In the third trimester, sexual interest can vary significantly. Fatigue might resurface as a challenge, and the growing abdomen can make intercourse feel uncomfortable. Some may experience unfounded fears that sexual activity could affect the baby or induce premature labor, though there's no evidence supporting these concerns. Medically, abstaining from intercourse and orgasm might be advised in cases of a history of premature labor, a placenta placed abnormally over the cervix, or ruptured amniotic membranes. If these conditions are absent, continuing sexual activity is generally safe until the onset of labor. Your healthcare provider will advise you on any circumstances requiring a halt to sexual relations.

Suggestions for Sex While Pregnant

- **Position:** Adjustments in positions for intercourse will need to be made as the abdomen grows. Often, the side-by-side position is most comfortable. Try new positions while keeping in mind comfort and pleasure.
- **Lubrication:** Because the vagina tends to be drier during pregnancy, lubricants for intercourse may enhance comfort.
- **Oral sex:** It is okay to have oral sex while pregnant. However, during pregnancy it is not safe to blow air into the vagina, as this may cause a life-threatening condition called air embolism.
- **Condoms:** Condoms are necessary to protect from the transmission of sexually transmitted infections (STIs) such as chlamydia, gonorrhoea, and HIV. It is important to continue using condoms if you risk exposure or are not in a mutually monogamous relationship.



Safety during Pregnancy

Tobacco/Smoking/Vaping

Smoking puts both mother and fetus at risk. When the mother smokes, the fetus is exposed to chemicals such as carbon monoxide and nicotine. Studies show that smoking during pregnancy can lead to complications like vaginal bleeding, miscarriage, premature delivery, neurologic impairment, stillbirth, and Sudden Infant Death Syndrome (SIDS). In addition, smoking reduces the oxygen available to the mother and fetus. Babies of mothers who smoke are likely to weigh less and be smaller. Infants and children who are exposed to environments where adults smoke can also have adverse effects.

We encourage you to avoid smoking (and secondhand smoke) as much as possible during your pregnancy. We can refer you and/or your partner to smoking cessation programs to help you quit.

Drugs/Medications

Drugs of any type (including illicit "street" drugs, prescriptions, or over-the-counter medications) may affect the foetus. Some may cause severe problems, while others may have no adverse effect. Review with your provider any medication you have been taking or plan to take to ensure that it is safe for you and your baby. If you have been taking medication regularly, check with your provider before stopping it, as the underlying condition treated by the medication may worsen upon sudden cessation. If you have been seeing a specialist for a particular condition, we will likely ask you to make a maintenance appointment with that physician during your pregnancy.

Some common medications are especially dangerous to your baby, such as:

- Accutane
- Thalidomide (Thalomid)
- Acitretin (Soriatane)

Exposure to Chemicals & Radiation

It is common to be exposed to chemicals in the house and at work. However, anything you breathe or come into contact with could enter your bloodstream and reach your baby. It may be helpful to ask for assistance when handling particular substances during the following activities:

- **Hair colour/dye:** Hair colour and permanents are considered to be low risk. If you are considering any hair treatments, be sure to have them performed in a well-ventilated area. Have someone else apply the dye to your hair and make sure your scalp is rinsed thoroughly. If possible, try to avoid use during the first trimester.
- **Cleaning:** There is little evidence to suggest that the use of everyday cleaning supplies causes significant harm. It is best to avoid certain cleaning products such as ammonia and bleach, especially in an area that is not well ventilated. Instead, try using alternative cleaning solutions such as vinegar or baking soda, or switch to products that do not contain harsh chemicals.



- **Gardening:** Chemicals frequently used in gardening, such as insecticides, weed killers, and fertilisers, should be avoided.

- **Painting:** Oil- and lead-based paints are particularly dangerous. Use caution when painting while pregnant and avoid using paint removers/strippers, varnish, shellac, and turpentine. It is also important not to work in or renovate an area where lead paint may be present. While painting, be sure to work in a well-ventilated area and avoid eating or drinking in the room where you are painting.

- **X-Rays:** Minimal exposure to x-rays during pregnancy is considered safe. X-rays are thought to pose at most a remote risk to the baby. Most x-ray exams are performed on the legs, arms, chest, head, and teeth, minimizing radiation exposure to the reproductive organs. Wearing a leaded apron or collar can protect against any scattered radiation.

Hot Tubs/Saunas

Maternal exposure to extremely hot temperatures has been shown to cause birth defects. While comfortably warm tub baths and showers are safe, hot tubs and saunas are not. Avoid using them and engaging in other activities that raise core body temperature (e.g., hot yoga) above 100° F.

Falls & Abuse

It is important that your body remains a safe space for your baby to grow in. We recommend that you avoid any activity that heightens your risk of falling or suffering abdominal trauma.

If you are being abused, during your pregnancy or otherwise, you are welcome to contact our office at any time or call the National Domestic Violence Hotline: 1-800-799-SAFE (7233).

Pets

- **Dogs:** Dogs pose little to no threat to pregnant women. However, there is concern if a dog jumps on the abdomen of a pregnant woman. Although the likelihood of developing complications from this is low, it is best to train your dog not to jump and to avoid jumpy dogs. Also, avoid walking dogs that tend to pull forcefully on their leashes, as this could lead to a fall.

- **Cats:** Cats can carry toxoplasma, a parasite that is transferred to humans through contact with cat faeces. Toxoplasma is most often found in outdoor cats but can be found in indoor cats as well. It is best to have someone else at home change litter boxes; however, if you must be the one to clean the litter be sure to wear protective gloves and wash hands thoroughly after.

- **Amphibians/Reptiles:** Reptiles and amphibians, such as iguanas, turtles, snakes, and lizards, can transmit salmonella to humans who are exposed to their faeces. Wash your hands after handling these pets, avoid having these pets around the kitchen/food preparation area, do not bathe them or clean the cage in the kitchen sink, and do not let them roam freely around the house.

- **Birds:** Birds are generally safe but can carry bacteria like salmonella and campylobacter. A visit to the veterinarian to determine the health status of your bird is helpful. Wash your hands after handling your pet and avoid being the one who cleans the cage if possible.



Infections during pregnancy

Infections may be minor and have no effect on the fetus (e.g., a cold) or they may cause a serious, life-threatening illness for the mother and/or fetus. If you believe you have been exposed, or have symptoms of infection, please feel free to call us.

We routinely screen pregnant women for exposure or immunity to the following diseases, as they may be particularly hazardous to a fetus:

- HIV/AIDS
- Syphilis
- Hepatitis B
- Rubella (German measles)
- Varicella (Chickenpox)
- Group B Streptococcus

Group B Streptococcus (GBS):

This is a common bacterium that usually colonizes the gastrointestinal tract and also the vagina/genitals in some people. It is estimated that up to 25% of pregnant women are carriers. While GBS generally does not cause any ill effects pregnant women, in rare cases, if a baby is exposed to the bacteria during labour and delivery, it can cause severe effects such as sepsis or meningitis. We routinely screen women for GBS at 34- 36 weeks gestation, and prophylactically treat women who are positive for the bacteria in labour with IV antibiotics to reduce the chance of neonatal infection

Other infections that may require careful monitoring, testing, or follow-up:

- **Listeria:** A serious infection caused by eating food contaminated with the bacterium *Listeria monocytogenes*. The disease primarily affects pregnant women, newborns, and adults with weakened immune systems. Symptoms of listeria include high fever, general malaise, and muscle aches. Call your provider if you are experiencing these symptoms. See "Foods to Limit" (page 8) for more information on potential contaminants.
- **Toxoplasmosis:** An infection caused by a parasite called *Toxoplasma gondii*. Of those who are infected, very few have symptoms because a healthy person's immune system usually keeps the parasite from causing illness. However, pregnant women and individuals who have compromised immune systems should exercise caution. Wear gloves and wash hands after gardening or handling soil, wear gloves or have someone else change the cat's litter box, cover outdoor sandboxes, and follow good food safety practices. Also see "Foods to Limit" (page 8).
- **Cytomegalovirus (CMV):** A common virus that, once in a person's body, can reactivate. CMV is passed through body fluids and can also be transmitted from mother to child during pregnancy. CMV symptoms include fever, sore throat, fatigue, and swollen glands. If you think you may have CMV, contact your care provider for more information.



- **Herpes:** Caused by the herpes simplex virus. A primary infection in pregnancy or having an outbreak close to the time of delivery can have negative effects on the fetus. Alert your health care provider if you or your partner have ever been exposed to genital herpes, you have ever had a herpes outbreak, or you have a new genital lesion during pregnancy.
- **Zika:** Zika virus infection during any trimester of pregnancy has been found to cause adverse birth outcomes such as pregnancy loss, microcephaly, and other brain and eye abnormalities. The virus spreads through infected mosquitoes, from a mother to fetus during pregnancy, and through sexual contact. However, it may also be spread through blood transfusion and other laboratory exposure. Symptoms of the virus include acute onset of fever, rash, joint pain, and conjunctivitis. Currently, there is no vaccine or treatment for Zika virus. It is advised that pregnant women DO NOT TRAVEL to areas where Zika outbreaks are documented.
- **COVID-19:** At this time, pregnant women are not considered a high-risk population for the novel coronavirus. However, pregnant women have changes in their bodies that may increase their risk of some infection, and pregnant people have had a higher risk of severe illness when infected with viruses from the same family as COVID-19 and other viral respiratory infections, such as influenza. We are currently following the CDC for the most up-to-date guidelines: www.cdc.gov/coronavirus/2019-ncov

Vaccines during Pregnancy

There are two vaccines that we recommend pregnant women get during each pregnancy:

- **Influenza Vaccine (Flu Vax):** Pregnant women are at greater risk of becoming severely ill from the flu due to decreased immunity and decreased lung capacity. It is recommended that pregnant women receive a flu shot during each flu season.

The FluMist nasal flu vaccine is NOT recommended in pregnancy as it contains live virus.

- **DPT Vaccine (Diphtheria, Pertussis, Tetanus):** We recommend that pregnant women receive the vaccine during each pregnancy, ideally between 20-34 weeks gestation. This helps ensure that antibodies are passed from mom to baby to help protect against whooping cough (pertussis) after the baby is born. Newborns cannot be vaccinated themselves for pertussis until several months of age. It is recommended that anyone who may have close contact with a newborn ensure that they are up to date on their Tdap vaccine.



Common Testing in Pregnancy

Genetic Testing

The purpose of genetic testing is to achieve early insight into a baby's development, attain more information, and screen for or diagnose a birth or genetic defect. Genetic testing also assists providers in caring for your pregnancy and determining if a baby may need special care immediately after birth. Early genetic testing can also give parents time to research/prepare for a baby who might have specific genetic problems, acquire early specialist care, make informed decisions about the method of becoming pregnant or about continuing an existing pregnancy. Even if you would not terminate a pregnancy that was found to be abnormal, many people undergo genetic testing for the knowledge that is acquired. **All genetic testing is optional.**

- **Genetic Counselling:** Genetic counselling involves meeting with a specially trained professional to find out more about your genetic makeup and how it may affect you and/or your future baby. The following are some situations in which you may want to consider genetic counselling:

- A family history of a genetic condition
- To learn more about genetic conditions/diseases related to certain ethnic groups
- To discuss abnormal test or ultrasound results during your pregnancy
- To discuss the effects of harmful substances and/or other exposures
- Infertility
- Birth defects in previous pregnancies
- Preparation for a healthy pregnancy

- **Genetic Screening Tests:** Genetic screening can tell you the chances that your baby has or will have a genetic disorder. There are several test options, including Carrier Screening, the First Trimester Screen, Non-Invasive Prenatal Screening, and Maternal Serum Alpha-Fetoprotein Screening.

- **What is it for?** Screens to see if you carry a specific gene that causes a specific inherited disorder
- **Who is it for?** People who are considering trying to conceive, newly pregnant, or wishing to get more information about the genes they could pass on to a child
- **When can it be done?** Any time prior to or during a pregnancy, and only needs to be done once
- **How is it done?** Blood test or saliva sample



Examples of diseases included in Carrier Screening:

Disease	Carrier Frequencies	Notes
Cystic Fibrosis (CF)	1 in 28 carries a mutation of CF.	It affects 1: 2500 babies
Spinal Muscular Atrophy (SMA)	1 in 40 carries one faulty copy of the SMA1 gene	SMA is the most common genetic cause of death in infants under 2 years of age.
Fragile X syndrome (FXS)	1;250 women and 1:800 men are carriers of FXC	1: 4000 males and 1: 8000 females FXS causes mild to severe intellectual disability.

The First Trimester Screen & Diagnostic Tests

The First Trimester Screen

Also called?

- FTS, NTD, NT, Nuchal Translucency + Blood Work

What is it for?

- Screening test to assess a mother's risk for having a baby with Trisomy 21 (Down syndrome), Trisomy 18 (Edwards Syndrome), and Trisomy 13 (Patau Syndrome)
- Optional testing for presence of Y-chromosome (Male sex chromosome)

Who is it for?

- Offered to all pregnant women
Particularly those at lower risk for genetic abnormalities (i.e., women under age 35)

When can it be done?

- Only during the first trimester

How is it done?

- Blood test (between 9 0/7 weeks- 13 6/7 weeks gestation)
- Ultrasound measurement (11 0/7 weeks- 13 6/7 weeks gestation)

Additional Information: First Trimester Screen

- The maternal blood test measures levels of specific biochemical hormones, including:
- Human chorionic gonadotropin (beta-hCG) and pregnancy-associated plasma protein A (PAPP-A).



- Beta-hCG is a hormone made by the placenta. High or low levels may be related to certain birth defects. PAPP-A is a protein in the blood. Low levels may be related to certain birth defects.
- The ultrasound detects the presence of a nasal bone and measures the amount of fluid accumulation behind the fetal neck
- Also called the nuchal translucency or NT ultrasound
- An increased NT can be indicative of genetic abnormalities, or other structural defects
- Absent nasal bone can be indicative of genetic abnormalities and other structural defects
- Additional screening can also be added to screen for presence of the Y-chromosome to determine fetal sex. (Note: this is usually at an increased cost.)
- The results of the ultrasound are combined with the results of the blood test and maternal age to give a risk assessment of having a baby with trisomy 21,13, or 18.
- These combined biochemical and biophysical markers yield very sensitive results, achieving a 93% detection rate at a 5% false positive rate for Down syndrome, and a 95% detection rate at a 0.3% false positive for Trisomy 18 and Trisomy 13. If the fetal nasal bone assessment is included, the detection rate for Down syndrome increases to 96% at a false positive rate of just 2%.

Example of First Trimester Screen Results:

Serum Markers	Value	MoM	Percentile		Biophysical Markers	Value	Delta/MoM	Percentile
Free Beta hCG	33.84 (ng/ml)	0.91	50		NT	2.1(mm)	0.47	75
PAPP-A	552 (mU/l)	0.29	2		Nasal Bone	Present	-	-
AFP	5.58 (IU/ml)	0.88	40		-	-	-	-
PIGF	17 (pg/ml)	0.61	20		-	-	-	-
Inhibin-A	366.54 (pg/ml)	0.72	30					

Risk Table	Cut-Off	Risk Before Screening	Risk After Screening	Result									
Down Syndrome	1 in 300	1 in 197	1 in 2,883	WITHIN RANGE									
Trisomy 18/13	1 in 150	1 in 361	1 in 796	WITHIN RANGE									
Down Syndrome	Decrease Risk Zone			Increased Risk Zone									
Before													
After													
Age:	20	22	24	26	28	30	32	34	36	38	40	42	44
Trisomy 18/13	Decreased Risk Zone						Increased Risk Zone						
Before													
After													



What if a screen comes back positive or indicates elevated risk?

If a screen comes back positive, you have the option of being referred for genetic counselling and of doing additional testing, including:

- Additional blood work for non-invasive prenatal testing (NIPT)
- Diagnostic testing, including chorionic villus sampling or amniocentesis

Non-Invasive Prenatal Screening

Also called?

- NIPS, non-invasive prenatal testing (NIPT), cell-free DNA (cfDNA), Prequel Screen

What is it for?

- Screening test to assess a mother's risk for having a baby with Trisomy 21 (Down syndrome), Trisomy 18 (Edwards Syndrome), and Trisomy 13 (Patau Syndrome)
- Optional screening for presence of and abnormalities of the sex chromosomes
- Optional screening for chromosomal microdeletions

Who is it for?

- Offered to all pregnant women, particularly those at higher risk for genetic abnormalities (i.e., women over age 35), or who have a personal or family history of chromosomal abnormalities
- Women with twin pregnancies and donor gametes
- Women who have had a previous positive First Trimester Screen

When can it be done?

- Any time during the pregnancy and as early as 10 0/7 weeks

How is it done?

- Maternal blood test any time after 10 weeks

Additional Information: Non-Invasive Prenatal Screening (Myriad PREQUEL Screen)

A blood sample is analysed in the lab to measure fragments of fetal DNA in the maternal bloodstream (cell-free DNA). It looks for abnormalities in the amount of DNA from chromosomes 21,18,13, and the sex chromosomes. It can also detect some forms of microdeletions, or missing parts of chromosomes.

Conditions Screened:

Trisomy Disorders	Sex Chromosomes Abnormalities
Trisomy 21 (Down Syndrome)	X (Turner Syndrome)
Trisomy 18 (Edwards Syndrome)	XXX (Trisomy X)
Trisomy 13 (Patau Syndrome)	XXY (Klinefelter Syndrome)
	XYY

Prequel screening is about 97-99% accurate, with a small chance of false positive and a very small (0.1%) chance of a false negative. Prequel screening includes individualized positive predictive values (chance of a true positive) and residual risk based on age.



Of note: If undergoing NIPS screening, we generally still recommend an early anatomy scan and nuchal translucency ultrasound at 12-13 weeks to ensure appropriate fetal growth, and also because an increased nuchal translucency can indicate some cardiac or other structural abnormalities.

Limitations to Non-Invasive Prenatal Screening: All non-invasive prenatal screens have limitations. A normal result reduces, but does not eliminate, the chance of chromosomal abnormalities. It is important to know that these tests are **NOT** diagnostic and that any positive results should be followed up by CVS or amniocentesis (diagnostic tests) for confirmation, as well as referral to a genetic counsellor.

Maternal Serum Alpha-Fetoprotein Screening

Also called?

- MSAFP, Alpha-fetoprotein, AFP

What is it for?

- Screening test for open neural tube defects, such as spina bifida or anencephaly, and other genetic defects

Who is it for?

- All pregnant women

When can it be done?

- Second trimester, usually between 15-20 weeks gestation

How is it done?

- Maternal blood draw

Additional Information: MSAFP

This blood test measures the level of alpha-fetoprotein in the mother's blood during pregnancy. AFP is a protein normally produced by the baby's liver and is present in the fluid surrounding the fetus (amniotic fluid) and crosses the placenta into the mother's blood.

An elevated AFP level can indicate open neural tube defects (abnormalities in the brain or spinal cord) such as spina bifida or anencephaly. It can also indicate problems with the fetal digestive system such as abdominal wall defects.

The MSAFP screening can also be combined with other hormone tests for patients who did not receive first trimester genetic screening. This is called the Quad Screen/Multiple Markers screen.

Abnormal screens are usually referred for detailed anatomy ultrasound, genetic counselling, and optional amniocentesis.



Diagnostic Testing

Prenatal diagnostic tests can tell you whether your baby has a genetic disorder. They offer more certainty than the screening tests discussed above. Diagnostic tests use cells from the fetus or the placenta in order to provide a diagnosis. There are two main diagnostic tests: Chorionic Villus Sampling and Amniocentesis.

Chorionic Villus Sampling (CVS)

What is it for?

- Diagnostic genetic testing in the first trimester

Who is it for?

- Any pregnant patient seeking first trimester diagnostic genetic testing
- Patients with high risk for genetic anomalies, or women over age 35
- Patients who receive abnormal genetic screening test results
- Parents who are both carriers for the same genetic disorder
- Patients who have had a previous pregnancy with a genetic anomaly
- Abnormal first trimester ultrasound results

When can it be done?

- First trimester between 10-13 weeks gestation

How is it done?

- A sample of tissue is taken from the placenta and sent to the lab for analysis

Additional Information: Chorionic Villus Sampling

This is a diagnostic test that is offered to all pregnant women; however, it is typically indicated for higher risk pregnancies. CVS is a test where a small piece of the placenta (called chorionic villus) is collected, and then tested to check for chromosomal or genetic disorders in the baby. It is completed between 10 and 13 weeks of pregnancy. The testing is done at the Maternal Fetal Medicine office.

Two different methods may be used to collect the sample. One method uses a tube guided through the cervix under sonographic guidance. The second method uses a needle placed through the abdomen and directed through the uterus and into the placenta with sonographic guidance. A similar sample is obtained with both methods. This is considered invasive testing.

Risks are small, but may include:

- Miscarriage (0.7-1.3%)
- Infection
- Bleeding
- Fetal limb defects
- Rupture of membranes



Amniocentesis

Also called?

- Amnio

What is it for?

- Diagnostic genetic testing in the second trimester
- Testing for other disorders of the fetus

Who is it for?

- Any pregnant patient seeking second trimester diagnostic genetic testing
- Those with high risk for genetic anomalies, or women over age 35
- Patients who receive abnormal genetic screening test results
- Those who receive abnormal CVS results
- Parents who are both carriers for the same genetic disorder
- Patients who have had a previous pregnancy with a genetic anomaly
- Abnormal first or second trimester ultrasound results
- Confirmation of neural tube defects

When can it be done?

- Second trimester, usually between 15-18 weeks gestation

How is it done?

A sample of amniotic fluid is taken and sent to the lab for analysis.

Additional Information: Amniocentesis

This is a diagnostic test that is offered to all pregnant women; however, it is typically indicated for higher risk pregnancies. Amniocentesis uses sonographic guidance to direct a needle into the fluid sac around the baby and collect a sample. The fluid is tested to measure certain levels of proteins, as well as the baby's chromosomes. The test can diagnose chromosomal disorders such as Down Syndrome (Trisomy 21) and other inherited genetic disorders. It can also diagnose issues like open neural tube defects. It may be performed as soon as 15 weeks gestation. This is considered an invasive test, and it is done at the Maternal Fetal Medicine office.

Risks are small, but may include:

- Fetal loss (rate of less than 1%)
- Fetal injury
- Leakage of amniotic fluid
- Infection
- Rupture of membranes



Routine Lab Investigations

1st visit labs:

- HIV, hepatitis B, immunity to rubella, varicella, syphilis
- Blood type and Rh status, antibody screen
- Assessment for anaemia, or haemoglobin disorders like sickle cell disease
- Swab or urine test for gonorrhoea/chlamydia
- Screening of urine to check for asymptomatic urinary tract infections
- Cervical screening test (CST) for cervical cancer if not up to date

26-28 week labs:

- Recheck blood levels for signs of anaemia
- Recheck for blood type/antibody testing
- Screening for gestational diabetes
- * Recheck for syphilis.

34-36 week labs:

- Recheck for signs of anaemia
- *Recheck for blood type/ antibody testing
- Swab for group B streptococcus (GBS)

Gestational Diabetes Screening/Testing

Gestational diabetes (or diabetes of pregnancy) is a condition of impaired glucose(sugar) metabolism. It occurs due to decreased insulin function in the setting of pregnancy hormones. It is a common condition during pregnancy and often does not have any symptoms. Gestational diabetes occurs in approximately 5-10% of pregnancies in Australia.

The main risks of gestational diabetes include:

- **Macrosomia** (i.e., large baby): This can make vaginal delivery more difficult and caesarean delivery more likely.
- **Placental Insufficiency.** This is a situation in which the placenta cannot adequately support the pregnancy.
- **Neonatal Hypoglycaemia:** This is a condition of low blood sugar during the baby's first hours of life.
- **Hypertensive disorders:** Women with gestational diabetes are more likely to develop high blood pressure in pregnancy.

2-Hour Glucose Tolerance Test (GTT)

- **YOU NEED TO BOOK WITH LAB IN ADVANCE as it takes 2 hours.**
- * **THIS IS A FASTING TEST.** Do not eat or drink anything except plain water for 8-10 hours before the test.
- Your fasting blood glucose level will be drawn before drinking a 75 gam glucose solution.
- You will have blood levels drawn at 1 hour and 2 hours, after finishing the drink.



Results: If the 2-hour test is abnormal, a diagnosis of gestational diabetes is made. Once diagnosed, a patient will be referred to our diabetes team for further evaluation and management. Specific counselling regarding diet/lifestyle modifications and increased fetal testing will be given.

Ultrasound/Sonography

A variety of ultrasounds will be done throughout the pregnancy, including the following:

- **Dating Ultrasound:** Usually done early in the first trimester to confirm gestational age with expected dating based on the last menstrual period, to confirm that the pregnancy is in the uterus, and to determine whether or not there is more than one fetus. Ultrasound measurements and dating are most accurate in the first trimester.
- **Nuchal Translucency Ultrasound (NT) / Early pregnancy anatomy ultrasound:** Done at approximately 12 weeks to measure the fluid level behind the baby's neck and to check on the growth of the baby.
- **Anatomy Scan (Level II) Ultrasound:** Detailed and comprehensive anatomy ultrasound of the baby to look at and measure all of the fetal parts and long bones and make sure that all of the appropriate structures are present. It evaluates for abnormalities, looks at the genitals, and can confirm fetal sex. It also looks at the location of the placenta and umbilical cord and the length of the cervix.
- **36-week Ultrasound:** Usually done to assess fetal growth and head presentation and evaluate the need for additional testing before 40 weeks.
- **40-week Ultrasound:** Usually done to check fetal well-being, evaluate amniotic fluid levels, and ensure that it is safe to continue the pregnancy. It also verifies that the placenta is continuing to function properly beyond the due date.

Other ultrasounds may be indicated more frequently for women who are over 35, who are carrying twins, or who need more frequent testing. These may include interval growth ultrasounds, testing for fetal well-being, evaluating amniotic fluid levels, and measuring cervical lengths.

CTG / Non-Stress Test (NST)

A Non-Stress Test may be performed during the third trimester to evaluate fetal well-being. During an NST, an external fetal heart rate monitor is placed on the abdomen to record the baby's heart rate, and a second monitor is placed on the mother to detect any uterine contractions or movements. This non-invasive test helps assess the baby's response to its own movements and helps determine the baby's overall health and well-being.

