***Prahran East Medical Centre***

**400 High St, Prahran 3181 P: 9510-8888 F: 9510-2666 E: info@prahraneastmc.com.au**

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Testing for Abnormal Recessive Genes and Chromosomes

About 4% of babies in Australia are born with significant abnormalities, and half of these are major. Some can be prevented with good nutrition and medical care. Most of the rest can be detected with prenatal diagnosis, comprising the routine ultrasounds and gene testing. Currently, government funding for gene testing is limited and out-of-pocket costs apply. These tests are described below and are considered optional.

1. **Genetic Carrier Screening** (cost approx $350).

Since 2019, all couples in Australia who are either planning a pregnancy or in early pregnancy are offered Genetic Carrier Screening. This is a test of the genetic make-up of cells in blood or saliva. It detects whether a chosen partner (typically the mother) is a carrier for so-called 'autosomal recessive' conditions that are serious, incurable and can begin in early childhood, such as Cystic Fibrosis (CF) or Fragile X. If she is found to be a carrier, her partner is tested for carrier status of that particular condition. Because each condition is quite uncommon (CF is the most common, with 1 in 25 people being carriers), the partner will usually be negative for any particular condition that might be found to be carried by the woman. But if he does carry that same gene, any pregnancy the couple has would have a 1 in 4 chance of resulting in a baby that suffers from the condition. Prenatal diagnosis would then be offered early in a pregnancy.

Although many couples are only offered testing for the three most common conditions (CF, Fragile X and Spinal Muscular Atrophy), there are around 300 conditions that can be tested for, all serious, incurable and often starting in early childhood. The cost of the test is similar for the three or all 300, so I recommend testing for the lot. The only downside is that the more conditions you look for, the more likely you are to find one, and it is more likely than not that carrier status to some condition will be found. Again, the partner will very likely be negative for the same rare gene. The partner test is usually much cheaper or free.

2. Chromosomal conditions: **Combined First Trimester Screen** ($100) or **NIPT** ($400)

If genes are the words in a genetic 'recipe book', chromosomes are the chapters. The most common and well known chromosomal abnormality is Trisomy 21, or Down Syndrome. More serious types that are incompatible with life beyond infancy include Trisomy 18. These can be tested for during the first trimester of pregnancy, starting at 10 weeks, by either of the two following methods:

a) **Combined First Trimester Screen (CFTS)**. Available since the early 2000s, this test combines data from a blood test for hormones at 10 weeks, an ultrasound to measure the skinfold thickness at the back of the baby's neck at 12 weeks, and the mother's age, to give an estimate of the risk of a major chromosomal abnormality. The accuracy of the test is such that it will detect at least 90% of affected pregnancies. Mostly, the result is 'low risk', and no further testing is recommended. When the result is 'increased risk', further testing is offered (see below).

b) **Non-Invasive Prenatal Testing (NIPT)**. The woman has a blood test at 10 weeks gestation, and sophisticated technology finds blood cells from the baby that have leaked into the mother's circulation (less than one in a million). These cells are directly tested for chromosomal abnormalities. This test is more than 99% effective at detecting chromosomal abnormalities.

With either CFTS or NIPT, if an abnormality is suspected, it needs to be confirmed by further testing to make a precise diagnosis. This is done by passing a fine needle, guided by ultrasound, into the placenta or the water around the baby. These tests are known as chorionic villus sampling and amniocentesis. Because they are invasive, they have a 1-2% risk of causing a miscarriage.

Further information is available at: ranzcog.edu.au/womens-health/patient-information-resources/prenatal-

screening-for-chromosomal-and-genetic-con; vcgs.org.au; and invitae.com.\

Healthy Eating in Pregnancy

There's a few things to keep in mind when choosing food during pregnancy. In the first trimester, if nausea is a problem, it's sometimes a case of any food is better than nothing. But aside from that, it's a good idea to keep the following issues in mind.

**The Good**

During the second and third trimesters (from 14 weeks), pregnant women and their babies need to eat more of certain **macronutrients**. The main ones are **protein, iron and calcium**. The healthiest sources of these foods are in legumes (like peas, beans, lentils and tofu), nuts and seeds, dark green leafy vegetables and whole grains. Lean meats, fish and dairy can also be eaten in small quantities. The other main nutrients can be ticked off by eating a variety of fresh fruit (especially berries) and vegetables, as well as spices. Regular exercise of at least 30 mins most days is also very important.

**The Bad**

A common mistake, however, is over eating, resulting in **excessive weight gain**. Weight gain of more than about 10kg during a pregnancy is weight that will persist beyond the pregnancy and can increase the risk of complications. Sweet sugary foods, refined carbohydrates like white bread, white rice, and non-wholemeal pasta eaten in large quantities, are common culprits, along with fatty dairy products like cheese and icecream.

There is mounting evidence that pregnant women should be advised to limit their intake of **pesticides and herbicides**, due to concerns about their impact on babies' neurological development. **Choosing organic food** options, where practical, is a good way to achieve this.

**Fish** has traditionally been recommended during pregnancy, because omega 3 fatty acids are required for healthy brain development. Unfortunately, fish these days come from polluted water, and women should restrict their fish intake, because of toxic heavy metals like mercury and microplastics, which are hormone disruptors. The healthiest way to get essential **omega 3s** is from the same source fish get them: algae. This is available in capsule form from some pharmacies and online (eg Deva Vegan Omega 3 from iHerb, 300mg per day). Ground **flaxseed** or flaxseed oil, 15g per day, takes care of the remaining omega requirements.

**The Ugly**

There a two important diet-related infections to be mindful of during pregnancy.

**Toxoplasmosis** is a virus that can cause inflammation of the brain (encephalitis) in unborn babies. It can be contracted from handling raw meat and not washing hands thoroughly afterwards, as well as cat litter.

**Listeria** is a bacteria that can cause a form of gastroenteritis that can infect the pregnancy and lead to miscarriage at any gestation. Fortunately, this only occurs very rarely. At-risk foods for listeria include deli meats like ham and salami, soft cheeses like feta, brie and Camembert, raw seafood, preprepared coleslaws, soft-serve icecream and raw egg white. Cooking kills the listeria bacteria.

**Supplements**

Other than omega 3 (see above), the only other supplement that is recommended is a pregnancy multivitamin like Elevit with Iodine, taken until 14 weeks gestation. This contains sufficient folate and other micronutrients to guard against deficiency-related birth defects. Elevit in particular has been shown to halve the risk of such defects compared to folate alone. Iodine is advised for Australian women because of the paucity of iodine in Australian soil, and the food that comes from it.

**Further reading**

These websites are worth checking out: YourFertility.org.au; nutritionfacts.org

Dr Martin Williams, MBBS, FRACGP, DRANZCOG