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Thanks to recent developments obstetricians at Mater Private are able to offer several options to expectant parents wanting to get information about their baby’s health as early as the first trimester. These screening tests can tell the likelihood of a baby having certain genetic conditions such as Down Syndrome, or even whether the baby is a boy or a girl.

Your obstetrician will discuss the risks and benefits of these tests in your unique situation, but it’s a good idea to have some understanding of what the options are. Combined first-trimester screenThe combined first-trimester screen has two parts: a blood test and an ultrasound. During the ultrasound, the baby’s length is measured to give a good estimate of its exact age, as well as the size of a small collection of fluid at the back of its neck called the nuchal translucency. All babies have this fluid, but the size of it can vary significantly. These figures are used along with measurements of different chemicals in the mother’s blood to calculate the chance of a baby having Down Syndrome, as well as two less common conditions called Patau and Edwards Syndrome. These conditions are known as trisomies and mean that the baby has an extra copy of a chromosome, or bundle of genetic information. The combined first-trimester screen is also known as a nuchal translucency test and is carried out between 11 and 13 weeks of a woman’s pregnancy, though the blood test can be done a little earlier. About 9 out of 10 babies with Down Syndrome will be identified by the combined first-trimester screen.

Non-invasive prenatal testing (NIPT)This newer test involves just a blood test. By about 10 weeks all mothers have a tiny proportion of their baby’s DNA or genetic material in their blood. The NIPT test involves taking a sample of the

mother’s blood and analysing the baby’s DNA. After this, the likelihood of the baby having genetic conditions involving extra copies of chromosomes such as Down, Edwards or Patau Syndromes can be calculated. Depending on the test the baby’s sex may also be determined as well as genetic conditions involving changes to the number of sex chromosomes such as Turner or Klinefelter Syndromes. This test may be more expensive than the combined first-trimester screen and can be completed from 10 weeks of pregnancy, depending on the exact test done.

About 99% of babies with Down Syndrome will be identified by non-invasive prenatal testing. It is important to realise that both of these tests are screening tests. This means that they identify women who are at a higher risk of having a baby with the conditions. They are not able to diagnose any of the syndromes. If you are identified to be at a higher risk of having a baby with one of these syndromes then your obstetrician will discuss further diagnostic tests which can be done at Mater Private.

These tests involve taking genetic material directly from the baby so that a syndrome can be definitively diagnosed.