Fact sheet **24**

PRENATAL TESTING OVERVIEW

This fact sheet talks about different ways of checking the health of a baby before birth, using a prenatal test.

IN SUMMARY

- Prenatal tests (tests in pregnancy) are a way of checking the health of a baby before birth
- There is no test that gives a 100% guarantee of a healthy baby
- Some tests give a highly accurate result about the health of the baby while others give an estimate or risk result
- Testing is always optional.

Every parent wants to have a healthy baby and for most, this will be the case. Sometimes, however, a baby will be born with a health or developmental condition. A health condition in the baby may happen for no apparent reason or because of a known risk factor.

Some of the reasons why the likelihood of a baby having a health concern is increased are:

- Where there is a close relative or a previous child with a health condition
- One or both parents of the baby has a health condition that may be passed on
- One or both parents of the baby are known 'carriers' of a particular gene variation so that the gene product is not made properly
- The mother is in her mid-30s or older (not necessarily her first pregnancy)
- There has been exposure to some chemical or other environmental factors during the pregnancy
- Where the parents are related by blood (e.g. first cousins).

WHAT ARE PRENATAL TESTS?

Prenatal tests can occur in a number of different ways and at certain stages of the pregnancy. In general, there are two main types of prenatal tests – a screening test and a diagnostic test.

Screening tests give a risk or estimate of the chance that a baby has a health condition. These tests do not generally look directly at a sample from the developing baby and are therefore very safe.

Included in the group of screening tests are ultrasounds, non-invasive prenatal testing (NIPT), first trimester screening and second trimester screening.

Each of these screening tests are talked about more in the diagram on the next page.

Diagnostic tests provide a more accurate result since they are testing the baby directly. Because of this, some of the tests may cause a loss of the baby in a small number of cases.

Included in the group of diagnostic tests are ultrasounds, chorionic villus sampling (CVS) and amniocentesis. These diagnostic tests are shown in the diagram and table on the next page.

WHAT IF I DO NOT WANT TO HAVE A PRENATAL TEST?

Testing in pregnancy is optional and should be discussed in full with your doctor, midwife or genetic counsellor. Making a decision to have a test or not is always up to you.

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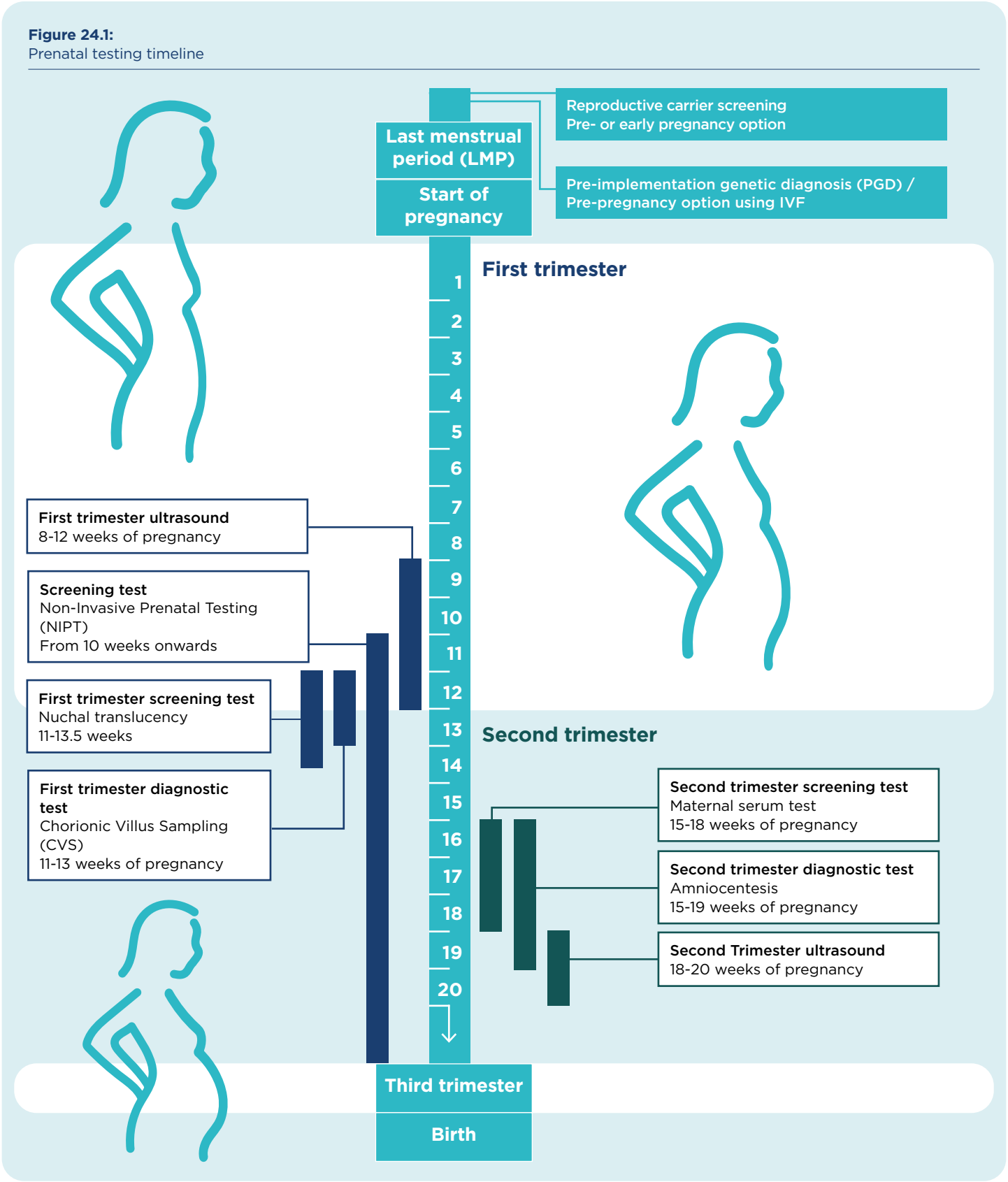
Figure 24.1:
Prenatal testing timeline



WHAT IF THE TEST SHOWS THE BABY HAS A HEALTH CONDITION?

When a baby is found to have a health condition before birth, it can be a very emotional and difficult time for the parents and family. You may be offered genetic counselling and given information about the health condition that has been identified, and supported as you understand its impact on your baby's health.

Depending on the stage of the pregnancy, you will be able to discuss options of planning for the birth of your baby or ending the pregnancy (having a termination of pregnancy). Whether you decide to continue or end a pregnancy, you will be offered support and information.

IF A TEST RESULT IS NORMAL, WILL MY BABY BE HEALTHY?

There is no test during pregnancy that can give a guarantee that a baby will have no health or developmental concerns. Even though diagnostic tests are more accurate than screening tests, they do not pick up every possible health condition.

PRENATAL TESTING TIMELINE

The prenatal testing timeline shows the different types of tests available at various stages of pregnancy. Each of these tests will give some information about the development of the baby.

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