PRENATAL TESTING – OVERVIEW

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Important points

- There are a number of different prenatal tests and procedures available to assess the development of the baby. Each has advantages, disadvantages and limitations
- There is no test that gives a 100% guarantee of a healthy baby. The tests give *some* information about the baby's health. They do not find *all* potential health problems
- Counselling before a test is done, will help the woman decide which test, if any, is best for the woman and the baby
- Each prenatal test is done at a certain time during the pregnancy starting at 8-10 weeks and going through to 20 weeks and include:
  - **Prenatal screening tests** that may identify a baby as being at an increased risk of having a particular problem. All pregnant women, regardless of their age or family health history, may choose to have one of these prenatal screening tests that include ultrasound; early pregnancy (first trimester) screening: nuchal translucency ultrasound with or without testing of the mother's blood; second trimester screening: testing of the mother's blood (maternal serum testing) (see Genetics Fact Sheets 17A and 17B)
  - **Prenatal diagnostic tests** that are used to see if the baby actually has a particular problem. Even if the test result is normal, however, the baby could still have some other problem. Prenatal diagnostic tests include ultrasound; chorionic villus sampling (CVS); amniocentesis; cordocentesis (see Genetics Fact Sheet 17C)
  - **Preimplantation genetic diagnosis (PGD)** is used to test the embryo created via *in vitro* fertilisation (IVF) therapy prior to implantation (see Genetics Fact Sheet 18)
- The consideration of **prenatal diagnostic testing** is indicated when
  - There is a close relative or a previous child with a serious condition
  - One of the partners in a couple has a serious condition that may be passed on to a baby
  - Both parents are 'carriers' of the same faulty gene
  - The mother is in her mid-30s or older (not necessarily her first pregnancy) as there is an increased risk for having a baby with chromosomal problems due to more or less than the usual number of chromosomes
  - There has been exposure to some chemical or other environmental agent
  - The results of screening tests such as ultrasound or first and second trimester screening tests have determined that the baby is at increased risk for a particular genetic condition in this pregnancy
- Prenatal **screening tests** are available to pregnant woman of all ages
- Genetic counselling is available if the baby is found to have an increased risk for a chromosome problem following a prenatal screening test, or is found to have a problem following diagnostic testing, and will provide an opportunity to discuss what the result means for the parents and the family, and provide support for their decision-making

Every couple wants to have a healthy baby. For most couples, this wish will come true. There are, however, some couples whose baby may have a serious physical or intellectual problem.

There are a number of different prenatal (meaning before birth) tests and procedures available to assess the development of the baby. Each has advantages, disadvantages and limitations. This information is for those who are

- Planning a pregnancy
- Already pregnant and want information about these tests

There is no test that can guarantee a baby will be healthy

The prenatal tests described in this series of Fact Sheets give *some* information about the baby's health. They do not identify *all* potential health problems. The woman can choose whether she wants any testing at all or which tests are best for her after talking to the doctor, midwife or genetic counsellor.

- Some people decide to have prenatal testing because they want to know if their unborn baby has a condition that causes serious physical and/or intellectual problems
- Some people decide not to have prenatal testing

The importance of counselling in association with prenatal testing

Counselling before any prenatal test is done, whether it is a screening test or a diagnostic test, is strongly recommended. It provides an opportunity to discuss:

- How and when the tests are done
- The advantages and disadvantages of each test
- Any risks to the baby that may result from each test
- Any further testing which the woman may be offered after she receives the result
- What this further testing will mean for the woman and the baby

Counselling before a test is done will help parents decide which test, if any, is best for the woman and the baby.

If the baby is found to have an increased risk for a chromosome problem following a prenatal screening test or is

found to have a problem following diagnostic testing, professional genetic counselling will provide an opportunity to discuss:

- What the result means for the parents and the family
- The options available to the parents at this time
- Whether they wish to have further testing if available
- The advantages and disadvantages of any further testing that is offered
- What course of action the woman and her partner wish to take

Support will be offered to the woman and her partner at this time, no matter what they decide to do.

What types of prenatal tests are available in Australia?

a) **Prenatal screening tests** may identify a baby as being at an increased risk of having a particular problem. Screening tests cannot determine if the baby definitely has a problem but may indicate that further testing needs to be considered (diagnostic testing). All pregnant women regardless of their age or family health history may choose to have one of these prenatal screening tests. Prenatal screening tests include:

- Ultrasound
- Early pregnancy (first trimester) screening: nuchal (pronounced *new-cal*) translucency ultrasound with or without testing of the mother's blood
- Second trimester screening: testing of the mother's blood (maternal serum testing)

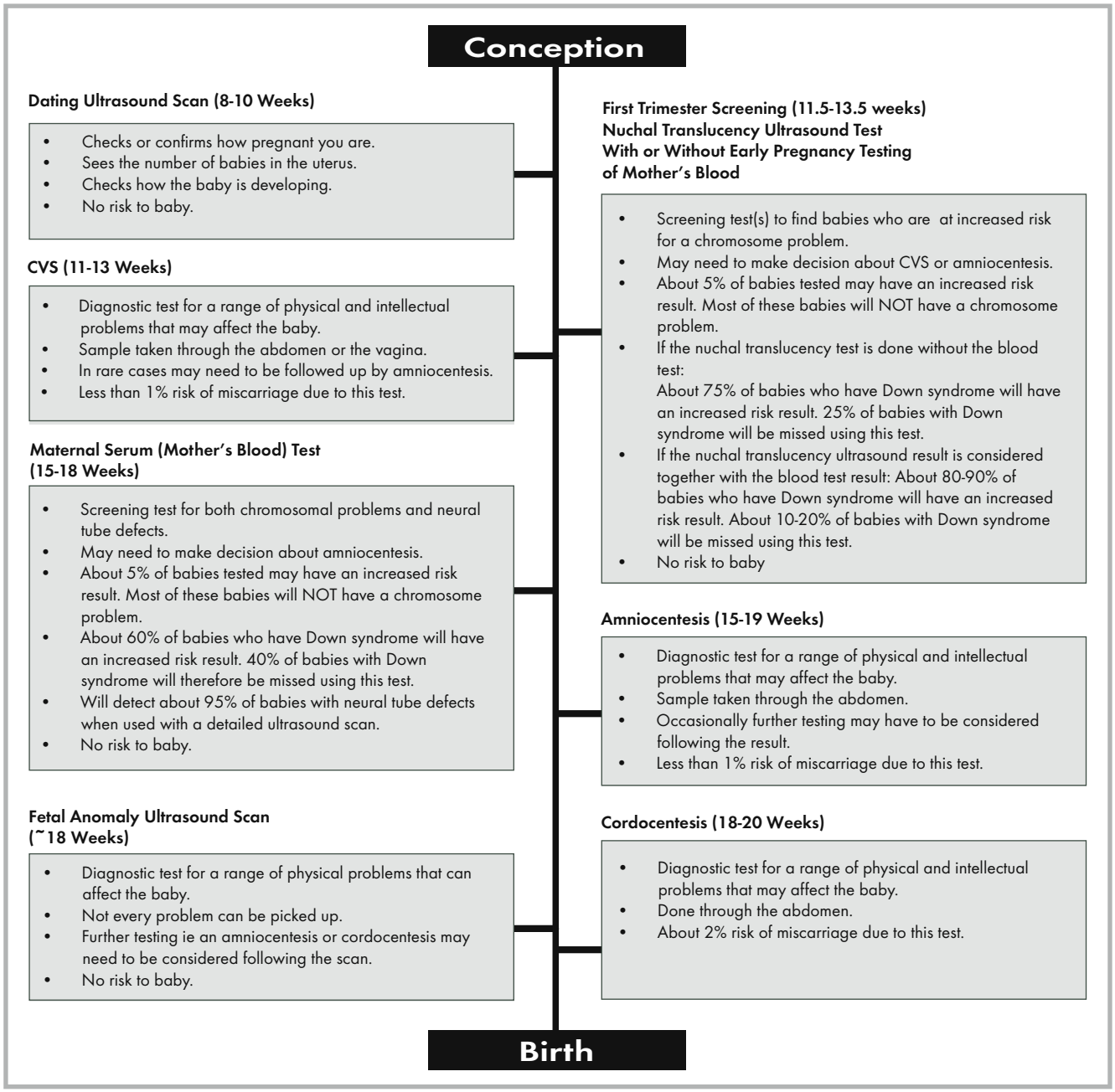
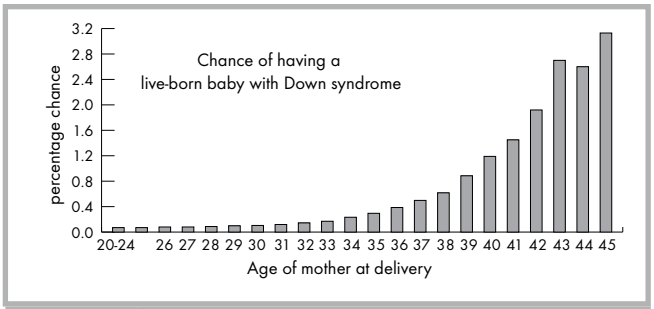
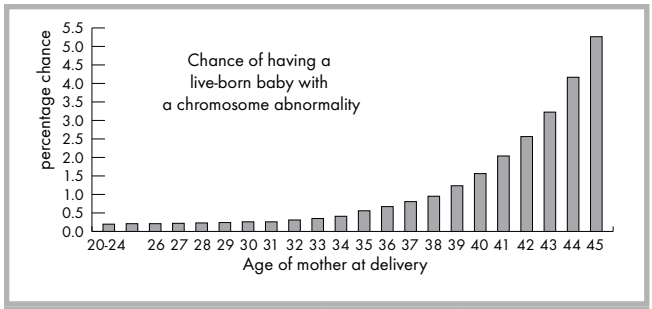


Figure 17.1. *Time line of prenatal testing including prenatal screening and diagnostic testing*



Age of mother at delivery	Chance of having a live-born baby with Down syndrome	Age of mother at delivery	Chance of having a live-born baby with Down syndrome
20-24 years	1 in 1411	35 years	1 in 338
25 years	1 in 1383	36 years	1 in 259
26 years	1 in 1187	37 years	1 in 201
27 years	1 in 1235	38 years	1 in 162
28 years	1 in 1147	39 years	1 in 113
29 years	1 in 1002	40 years	1 in 84
30 years	1 in 959	41 years	1 in 69
31 years	1 in 837	42 years	1 in 52
32 years	1 in 695	43 years	1 in 37
33 years	1 in 589	44 years	1 in 38
34 years	1 in 430	45 years	1 in 32

Figure 17.2: *Chance of having a live-born baby with Down syndrome (trisomy 21) according to the mother's age at the time of delivery of the baby. Source: Morris JK, Mutton DE, and Alberman E (2002). Revised estimates of maternal age specific live birth prevalence of Down syndrome. Journal of Medical Screening. 9,2-6*



Age of mother at delivery	Chance of having a live-born baby with a chromosomal abnormality	Age of mother at delivery	Chance of having a live-born baby with a chromosomal abnormality
20-24 years	1 in 506	35 years	1 in 179
25 years	1 in 476	36 years	1 in 149
26 years	1 in 476	37 years	1 in 124
27 years	1 in 455	38 years	1 in 105
28 years	1 in 435	39 years	1 in 81
29 years	1 in 417	40 years	1 in 64
30 years	1 in 385	41 years	1 in 49
31 years	1 in 385	42 years	1 in 39
32 years	1 in 323	43 years	1 in 31
33 years	1 in 286	44 years	1 in 24
34 years	1 in 244	45 years	1 in 19

Figure 17.3: *Chance of having a live-born baby with any chromosomal abnormality according to the mother's age at delivery. Source: Hook EB (1981). Rates of chromosomal abnormalities. Obs Gyn 58, 282-285.*

b) **Prenatal diagnostic tests** are tests done to see if the baby actually has a particular problem. Even if the test result is normal, however, the baby could still have some other problem. Prenatal diagnostic tests include:
   - Ultrasound
   - Chorionic villus sampling (usually simply called CVS)
   - Amniocentesis (pronounced *am-nee-o-cent-ee-sis)*
   - Cordocentesis

c) **Preimplantation genetic diagnosis (PGD)** that tests the embryo created with the use of *in vitro* fertilisation (IVF) therapy prior to implantation (see Genetics Fact Sheet 18)

When are prenatal tests done during pregnancy?

The different prenatal tests are only able to be done at certain times during the pregnancy starting at 8-10 weeks and going through to 20 weeks. A timeline for testing is shown in *Figure 17.1*.

What are the reasons for considering having a prenatal test?

Prenatal **screening tests** are available to pregnant women of all ages and, in particular when:

- There is a close relative or a previous child with a serious genetic condition (see Genetics Fact Sheet 2)
- One of the partners in a couple has a serious condition that may be passed on to a baby
- One or both parents are known 'carriers' of a particular faulty gene (see Genetics Fact Sheet 1)
- The mother is in her mid-30s or older (not necessarily her first pregnancy) as there is an increased risk for having a baby with chromosomal problems due to more or less than the usual number of chromosomes; eg. Down syndrome (see *Figures 17.2* and *17.3* and Genetics Fact Sheet 6)
- There has been exposure to some chemical or other environmental agent

- The results of screening tests such as ultrasound or first or second trimester screening may determine that the baby is at increased risk for a particular genetic condition in the pregnancy

This Genetics Fact Sheet provides an overview of all of the prenatal screening and diagnostic testing that is available in Australia, although access across the country is variable. Detailed information on each of the tests described above is provided in

- Genetics Fact Sheet 17A – Ultrasound
- Genetics Fact Sheet 17B – First and second trimester screening
- Genetics Fact Sheet 17C – CVS and Amniocentesis

Other Genetics Fact Sheets referred to in this Fact Sheet: 1, 2, 6, 17A, 17B, 17C,18

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Barlow-Stewart K, Emery J, Metcalfe S. (2007).Testing and pregnancy. In: *Genetics in Family Medicine: the Australian Handbook for General Practitioners*. Biotechnology Australia, Commonwealth Department of Industry, Tourism and Resources

Gardener, RJ and Sutherland GR. (2004). *Chromosome abnormalities and genetic counselling*, New York: Oxford University Press

Hook EB. (1981). Rates of chromosomal abnormalities. *Obs Gyn* 58 282-285

Morris JK, Mutton DE and Alberman E. (2002). Revised estimates of maternal age specific live birth prevalence of Down's syndrome. *Journal of Medical Screening*. 9, 2-6

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Author/s: A/Prof Kristine Barlow-Stewart and Mona Saleh

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