

## What if the further tests show your baby has Down syndrome, trisomy 18 or an open neural tube defect?

- Your doctor will explain the test results and explain all your options to you. Knowing this information may help to plan your baby's delivery. Some women may choose not to continue the pregnancy. You will also have the chance to learn about the help that exists for children with Down syndrome, trisomy 18 and neural tube defects.

### Open neural tube defects

Open neural tube defects happen when the brain or spinal cord does not develop properly. Between one and two babies out of 1,000 are born with open neural tube defects. There are two kinds of open neural tube defects: spina bifida and anencephaly. The chance of having a baby with an open neural tube defect does not depend on the age of the mother.

#### Spina bifida

In spina bifida, the bones of the spine are not fully joined. There may also be damage to the nervous system. Spina bifida causes physical and sometimes developmental disabilities. There is no way to predict how severe these effects will be. There is no cure, but treatment and support can help people with spina bifida.

#### Anencephaly

When a baby has anencephaly, the brain and skull do not grow completely. A baby with anencephaly will always die shortly after birth.

## Chromosome abnormalities

Chromosomes are carriers of genetic information. Down syndrome and trisomy 18 are chromosome abnormalities.

### Down syndrome

People with Down syndrome possess a wide range of abilities. All people with Down syndrome have a developmental disability. Some have added conditions that affect their health. There is no cure, but treatment and support can help people with Down syndrome.

Any woman at any age can have a baby with Down syndrome. About one in 800 babies is born with Down syndrome. The chance of having a baby with Down syndrome increases with a woman's age.

### Trisomy 18

Trisomy 18 is associated with many physical abnormalities. Most infants with trisomy 18 die by one year of age.

About one in 8,000 babies is born with trisomy 18. As with Down syndrome, the chance of trisomy 18 increases with a woman's age.

**If you have questions,  
speak to your doctor.**

# A Blood Test for Pregnant Women



## Prenatal Screening

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# Prenatal screening is a blood test for pregnant women that can provide some information about the health of your baby.

## What is prenatal maternal serum screening?

- A blood test all pregnant women can have as part of their prenatal care.
- The results of the blood test can tell you about the chance of having a baby with spinal defects (open neural tube defects) and some chromosome abnormalities (Down syndrome, trisomy 18). These conditions are not common. They are described later in this pamphlet.
- It is your choice whether to have prenatal screening. It will be done only if you want it.

## How is prenatal maternal serum screening done?

- A small amount of blood is taken from your arm. First trimester screening is done during your first trimester between 11 to 13 weeks of pregnancy. The second phase of prenatal screening is done during your second trimester between 15 and 20 weeks of pregnancy. The best time for the second trimester blood test is between 16 and 17 weeks.
- The amount of certain substances in your blood will be measured. These substances are made by the growing baby and the mother during pregnancy and are found in every pregnant woman's blood.

- The age of your baby must be known in order to interpret the blood test results. An ultrasound may be done at about the same time as the blood test to help check the age of the baby.

## What can this screening test result tell you?

- The screening test can tell you about the chance of there being certain abnormalities of the unborn baby.
- If the results of the blood test show that the levels of the substances do NOT follow the common pattern, your baby may have a higher chance of having a spinal defect or a chromosome abnormality. This result is called a risk above cut-off.
- Most women with a risk above cut-off have babies who do not have these conditions. Further tests will be needed to show if the baby does have one of these conditions. Talk with your doctor about further tests. Make sure all your questions are answered.
- If the results of the blood test show that the levels of the substances follow the common pattern, the chance of your baby having one of these conditions is low. This result is called a risk below cut-off.
- Ninety per cent of women will have risk below cut-off. However, even if the result is

a risk below cut-off, prenatal screening can sometimes miss Down syndrome, trisomy 18 or an open neural tube defect.

## What if the result is risk above cut-off?

- You can choose to have more tests to find out if your baby does have one of these conditions. You will be given choices about other tests.
- If there is an increased risk for Down syndrome or trisomy 18, you will be offered amniocentesis. During amniocentesis, a small amount of the fluid around your baby is taken and tested for Down syndrome or trisomy 18. Amniocentesis will cause a miscarriage in about one out of 200 women.
- If there is an increased risk for an open neural tube defect, you will be offered a detailed ultrasound. At the ultrasound, the head and back of your baby will be looked at. But an ultrasound cannot detect all cases of open neural tube defects. Amniocentesis might be offered when cases of open neural tube defects cannot be confirmed or ruled out with ultrasound.
- Most times the results of the ultrasound or amniocentesis will show that your baby does not have Down syndrome, trisomy 18 or an open neural tube defect.