



**RUH**

## Patient information: First trimester screening clinic



**This leaflet contains useful information about Down's Syndrome screening in the first weeks of your pregnancy and the choices available to you**

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## Contents

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<b>11-14 week Ultrasound Scan</b>	<b>3</b>
What is Ultrasound	3
Is it safe?	3
Why do I need to have an ultrasound scan?	3
Can I bring anyone with me to the scan?	4
What happens if there is a problem with my scan?	4
Preparation for your ultrasound scan	4
<b>Testing for Down's Syndrome</b>	<b>4</b>
What is Down's Syndrome?	4
What causes Down's Syndrome?	5
Should I have the test for Down's Syndrome?	5
Will the tests tell me for certain if my baby has Down's Syndrome?	5
Combined first trimester screening test	6
Second trimester screening	7
What is a high risk (screen positive) result?	7
What is a low risk (screen negative) result?	8
How long does it take to get the result of my screening test?	7
What happens if I have a high risk (screen positive) result?	8
Diagnostics tests for Down's Syndrome	9
Are these procedures safe?	9
What are the possible results from diagnostic tests?	9
What happens if my baby has Down's Syndrome?	9
<b>More information about screening</b>	<b>10</b>
<b>Your first trimester screening choices</b>	<b>11</b>

**Please read this booklet prior to attending the first trimester screening clinic and fill out the choices form on page 11.**



Choosing which tests to have in your pregnancy is an important decision for you and your baby.

This booklet gives you some information about Down's Syndrome and about testing, so you can decide whether or not to have the tests.

Please be aware that if any abnormalities are found on the early scan they will be discussed with you at the time of the scan, and referrals to a consultant will be made as necessary.

## 11-14 week ultrasound scan

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### What is ultrasound?

Ultrasound is high frequency sound waves that the human ear cannot hear. The sound waves produce echoes and these are used to create an image of your baby.

### Is it safe?

Ultrasound has been used for 30 years for monitoring pregnancies. So far the evidence has been reassuring that ultrasound is safe for mother and baby. However, we think it is wise to scan only when there is a good reason and to use the minimum amount of sound waves.

### Why do I need to have an ultrasound scan?

There are a number of reasons why we offer an ultrasound scan to all women between 11-14 weeks of pregnancy:

- To see the baby's heart beating
- Check whether there is just one baby
- Confirm how many weeks pregnant you are by measuring the baby





- Check there are no major abnormalities
- Offer screening for Down's syndrome

### Can I bring anyone with me to the scan?

Your partner or close friend is welcome to accompany you. Children are generally not encouraged to attend ultrasound examinations as this is a screening examination and they may distract you or our staff.

### What happens if there is a problem with my scan?

Sometimes it is not possible to check all the baby's anatomy especially if your dates are less than expected or your baby is lying in an awkward position. We will then offer you a repeat scan. Please do not worry if a re-scan is arranged for this reason.

If the person performing your scan is concerned about anything revealed during your scan, this will be explained to you. Another appointment will be offered to discuss the concerns and further tests may also be offered.

### Preparation for your ultrasound scan

- Please attend with a comfortably full bladder
- Please remove belly button jewellery

This booklet raises the question of possible problems being detected by ultrasound scan but please keep in mind that the majority of babies are perfectly healthy when they are born.

## Testing for Down's Syndrome

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### What is Down's Syndrome?

Down's Syndrome was first described by John Langdon Down



and the word ‘syndrome’ means a collection of characteristic features. People with Down’s Syndrome have learning difficulties but some are more severely affected than others. There are also physical problems associated with the condition; for example 40% will have a heart abnormality.

### **What causes Down’s Syndrome?**

Inside the cells of our bodies there are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. Most people have 23 pairs of chromosomes in each of their cells. When our bodies produce the cells needed to make babies, the chromosome pairs divide and rearrange themselves.

Sometimes these pairs of chromosomes accidentally do not divide correctly, and this can cause the baby’s cells to have an extra copy of chromosome 21. This causes Down’s Syndrome (also called Trisomy 21). The extra chromosome cannot be removed from the cells, so there is no cure for the condition.

### **Should I have the test for Down’s Syndrome?**

Only you can decide that. Some women want to find out if their baby has Down’s Syndrome, and some do not. Information about the tests and how they work can help you make up your mind.

### **Will the tests tell me for certain if my baby has Down’s Syndrome?**

We begin by offering all women a test that carries no risk of miscarriage. This type of test is called a screening test. Screening tests do not give a definite answer, but they do help us to tell which babies have an increased risk of having Down’s Syndrome. We then offer diagnostic tests to the women with an increased risk. These are tests that give definite information but they do carry a small risk of miscarriage. This is why we only





offer a diagnostic test to women who have been found to have an increased risk.

### **Combined first trimester screening test**

At this clinic you can choose to have a screening test for Down's Syndrome that involves a scan and blood test. This test can be taken from 11 weeks and 2 days up to 14 weeks and 1 day of pregnancy.

All babies have a very thin film of fluid under the skin at the back of their neck (nuchal area) at this stage of pregnancy. Provided your baby is lying in a suitable position, we can measure this as part of the scan and this is known as the nuchal translucency measurement.

If the nuchal translucency measurement is above 3.5mm, it is considered abnormal and will be discussed with you at the time of the scan, even if you choose not to have Down's Syndrome screening.

A blood sample is taken from the mother's arm during the same appointment as the scan (providing you are between 11 weeks and 2 days and 14 weeks and 1 day pregnant). The blood test measures the amount of some substances that are found naturally in the mother's blood. These substances have passed to the mother from the baby. If a baby has Down's Syndrome, different amounts of these substances can sometimes be found in the mother's blood.

The result of the blood test together with the mother's age, weight, and stage of pregnancy, (worked out by the scan) are combined with the nuchal translucency measurement, to work out the chance of your baby having Down's Syndrome. This is the combined test and the result is given as a risk figure. A risk can also be calculated for twin pregnancies.





## Second trimester screening

If you wish to have Down's Syndrome screening and your pregnancy is 14 weeks and over when you come to the first trimester screening clinic, you will be offered second trimester screening. This is a blood test and can be carried out between 14 weeks and two days – 20 weeks of pregnancy, and can be arranged with your midwife. The result is given as a risk figure.

### What is a high risk (screen positive) result?

Both the screening tests for Down's Syndrome give results in the form of, 'one in ...'; for example 'one in 100' (1%) or 'one in 1000' (0.1%). These numbers tell us how likely it is that the baby has Down's Syndrome. For example, the result 'one in 100' means that there is one chance in 100 that the baby has Down's Syndrome. The result 'one in 1000' means that there is one chance in 1000 that the baby has the condition. It is important to understand that as the second number in the result gets bigger, Down's Syndrome gets less likely.

If your screening result is above the national cut-off figure, we will offer you further tests. This is known as a high risk (screen positive) result.

### What is a low risk (screen negative) result?

If the screening test shows the risk of your baby having Down's Syndrome is lower than the national cut-off figure, this is known as having a low risk (screen negative) result and we will not offer you further tests. Most screening test results fall into this category.

It is important to understand that a low risk (screen negative) result means exactly that. It does not mean that there is no risk at all that your baby has Down's Syndrome, just that it is unlikely. There is still a small risk because some babies with Down's Syndrome are not detected by screening tests. Overall, about



a quarter of babies with Down's Syndrome are not detected by screening tests.

It is very important to remember that screening tests fit into a two-stage process, so if you get a high risk (screen positive) result, it means we will offer you more information and tests. It does not mean that your baby definitely has Down's Syndrome.

## How long does it take to get the result of my screening test?

### First Trimester Screening

The combined screening test result takes 2-3 working days.

### Second Trimester Screening

The second trimester screen result takes between 7-10 days. When the results are calculated the high risk (screen positive) results are telephoned to the mother, so it's very important that we have a contact telephone number. Low risk (screen negative) results will be sent out in the post. If you have not received your results within 10 days, please contact your named midwife.

## What happens if I have a high risk (screen positive) result?

If the result of the screening test shows the chance of your baby having Down's Syndrome is greater than the national cut off risk, we will offer you a diagnostic test. Overall about one in 33 (3%) women screened have a high risk (screen positive) result and are offered a diagnostic test.

- **You can decide not to have a diagnostic test**  
This carries no risk to your pregnancy but may leave some uncertainty until after your baby is born.
- **You can decide to have a diagnostic test**  
This will give you a definite result but carries a small risk of miscarriage.

You need to think about what you would do if you found yourself in this position. If you would not be happy with either of the above





options, you need to consider carefully whether the screening process is the right choice for you.

These screening tests may occasionally suggest a higher risk for a condition other than Down's Syndrome.

## Diagnostic tests for Down's Syndrome

There are two diagnostic tests offered for Down's Syndrome:

- Chorionic Villus Sampling (CVS)
- Amniocentesis (amnio)

For further information, please request individual leaflets from the First Trimester Screening Clinic.

### Are these procedures safe?

These procedures do carry some risk, and this is why we don't offer them to everybody. For every 100 women who have amniocentesis, one will miscarry. And for every 100 women who have CVS, one or two will miscarry.

### What are the possible results from diagnostic tests?

- **Your baby does not have Down's Syndrome**  
This is the most common result.
- **No or unclear result**  
Very rarely a CVS or amniocentesis does not produce a result because the cells do not grow, or the results are not clear. If this happens, further tests may be offered.
- **Your baby has Down's Syndrome**  
A very small number of women will get this result. It is not possible to tell how affected your baby will be.

### What happens if my baby has Down's Syndrome?

There are then three options – it is entirely your decision which one you choose:

- Some parents will decide to continue with the pregnancy,





make plans and prepare for any extra challenges they might face bringing up a child with Down's Syndrome

- Some parents may feel that they are unable to bring up their child themselves and may want to consider adoption as an option
- Some parents decide they do not want to continue with the pregnancy and will choose to have a termination.

**You have the opportunity to discuss your results with healthcare professionals. You will have time to decide what you are going to do and will be supported by your midwife, your obstetrician and your GP in your decision.**

## More information about screening

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You can get more information about screening from:

### **National Fetal Anomaly Screening Programme**

- Website: [www.screening.nhs.uk/fetalanomaly](http://www.screening.nhs.uk/fetalanomaly)

### **Antenatal Results and Choices**

- Website: [www.arc-uk.org](http://www.arc-uk.org)  
Helpline: 0207 631 0285

You can get more information about Down's Syndrome from the following organisations:

### **Contact a Family**

- Website: [www.cafamily.org.uk](http://www.cafamily.org.uk)  
Helpline: 0808 808 3555 (a free helpline for parents and families and is open from 10am to 4pm, Monday to Friday)

### **Down's Syndrome Association**

- Website: [www.dsa-uk.com.frameset.htm](http://www.dsa-uk.com.frameset.htm)  
Phone: 020 8682 4001



## Your First Trimester Screening Choices

Please fill in this form before attending the clinic.

Name: .....

Date of Birth: .....

NHS No: .....

Screening tests	Yes I would like to have this test	No I do not want this test	I don't know. I would like more information
Ultrasound scan			
Down's Syndrome screening			

I have read the information booklet and I understand the reasons for these tests and agree to this form being retained for hospital use.

Signed:..... Date:.....



The UK National Screening Committee has provided some of the information in this leaflet.

### Contact details:

First Trimester Screening Clinic  
D7, Princess Anne Wing, Royal United Hospital  
Combe Park, Weston, Bath BA1 3NG  
Phone: 01225 825414  
Monday to Friday 9am-4pm.

