

Bolton antenatal screening laboratory



**A guide to screening for
Down's, Edwards' and
Patau's syndromes**



Summer 2024

Version 5

This version supersedes version 4

Contact Details

For all antenatal screening service enquiries please contact Karina Hambridge or Trine Nielsen:

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Laboratory opening hours are from 8.45am – 5.00pm

The antenatal screening laboratory

The Department of Laboratory Medicine at Bolton performs serum antenatal screening for Down's, Edwards' and Patau's syndrome as part of the fetal anomaly screening programme (FASP) offered to all pregnant women in England.

The service covers over 70 clinics throughout Greater Manchester, Cheshire & Merseyside, Lancashire, North Midlands and Scotland, processing over 55,000 samples for Down's, Edwards' and Patau's syndromes chance calculations per annum.

The laboratory is run by a team of dedicated clinical and biomedical scientists and support staff.

Antenatal screening for chromosomal abnormalities

Down's (trisomy 21), Edwards' (trisomy 18) and Patau's (trisomy 13) syndromes arise from extra copies of chromosomes in the cells of the fetus. Screening for Down's (T21) syndrome is offered to all women between 10⁺⁰ and 20⁺⁰ weeks of pregnancy. First trimester screening is the UK National Screening Committee (NSC) recommended pathway¹. However, women who are unable to participate during the first trimester should be offered screening in the second trimester. Currently screening for Edwards' (T18) and Patau's (T13) syndromes is only available in the first trimester and a single chance for both of these conditions together is generated.

Screening should be carried out only once.

Antenatal screening

The laboratory provides a screening programme for Down's, Edwards' and Patau's syndromes that consists of:

1. First trimester combined testing (10⁺⁰ – 14⁺¹ weeks)

Chance for Down's, Edwards' and Patau's syndromes are calculated using the nuchal translucency (NT) ultrasound measurement combined with two biochemical markers:

- Pregnancy associated plasma protein (PAPP-A)
- Free-beta human chorionic gonadotrophin (β hCG)

2. Second trimester quadruple testing (14⁺² – 20⁺⁰ weeks)

Quadruple testing for Down's syndrome should only be offered to women who have been unable to participate in screening during the first trimester¹. The chance is calculated using the gestational age of the fetus combined with four biochemical markers:

- Inhibin-A
- Alpha-feto protein (AFP)
- Free-beta human chorionic gonadotrophin (β hCG) or total β hCG
- Unconjugated oestriol (uE3)

Biochemical & ultrasound measurements

The biochemical and ultrasound markers used in first and second trimester screening are described in the table below.

Marker	Trimester	Description	Changes in normal pregnancy
Nuchal translucency (NT)	First	Sonographic appearance of a collection of fluid under the skin behind the fetal neck.	Gradual increase in NT during the first trimester.
Pregnancy associated plasma protein (PAPP-A)	First	Produced by the placenta. Involved in the regulation of fetal-placental growth.	Blood concentration increases with gestational age.
Free-beta human chorionic gonadotrophin (βhCG)/or total HCG	First and second	Produced by the placenta. Important for the development of the fetal-placental unit.	Blood concentration increases in the early weeks of pregnancy, then begins to decrease after 10 weeks gestation.
Alpha-feto Protein (AFP)	Second	Produced by the fetal yolk sac and liver, but does not appear to be required for normal fetal development.	Blood concentration increases with gestational age.
Inhibin-A	Second	Produced by the ovary and placenta. Acts on the pituitary gland to prevent ovulation during pregnancy.	Blood concentrations remain stable throughout the second trimester.
Unconjugated oestriol (uE3)	Second	Steroid hormone, synthesised by fetal adrenals, liver and placenta. The major oestrogen of pregnancy.	Blood concentration increases with gestational age.

Chance calculations and use of multiples of the median (MoM)

Blood concentrations of biochemical markers change throughout pregnancy. For this reason results are divided by the median concentration value of unaffected pregnancies at the same gestational age and expressed as a multiple of the median (MoM). By definition the median MoM for unaffected pregnancies is 1.0. The MoMs are then used to calculate the chance of a pregnancy affected by Down's, Edwards' or Patau's syndromes.

The use of MoMs allows us to easily identify women with higher (MoM >1.0) and lower (MoM <1.0) marker results. Whilst they should not be interpreted in isolation of a chance calculation, typical patterns in affected pregnancies are as follows:-

First trimester screening (Down's syndrome)		First trimester screening (Edwards' and Patau's syndrome)		Second trimester screening (Down's syndrome only)	
Marker	MoM	Marker	MoM	Marker	MoM
NT	Increased	NT	Increased	Inhibin-A	Increased
PAPP-A	Decreased	PAPP-A	Decreased	AFP	Decreased
β HCG	Increased	β hCG	Decreased	β hCG	Increased
				uE3	Decreased

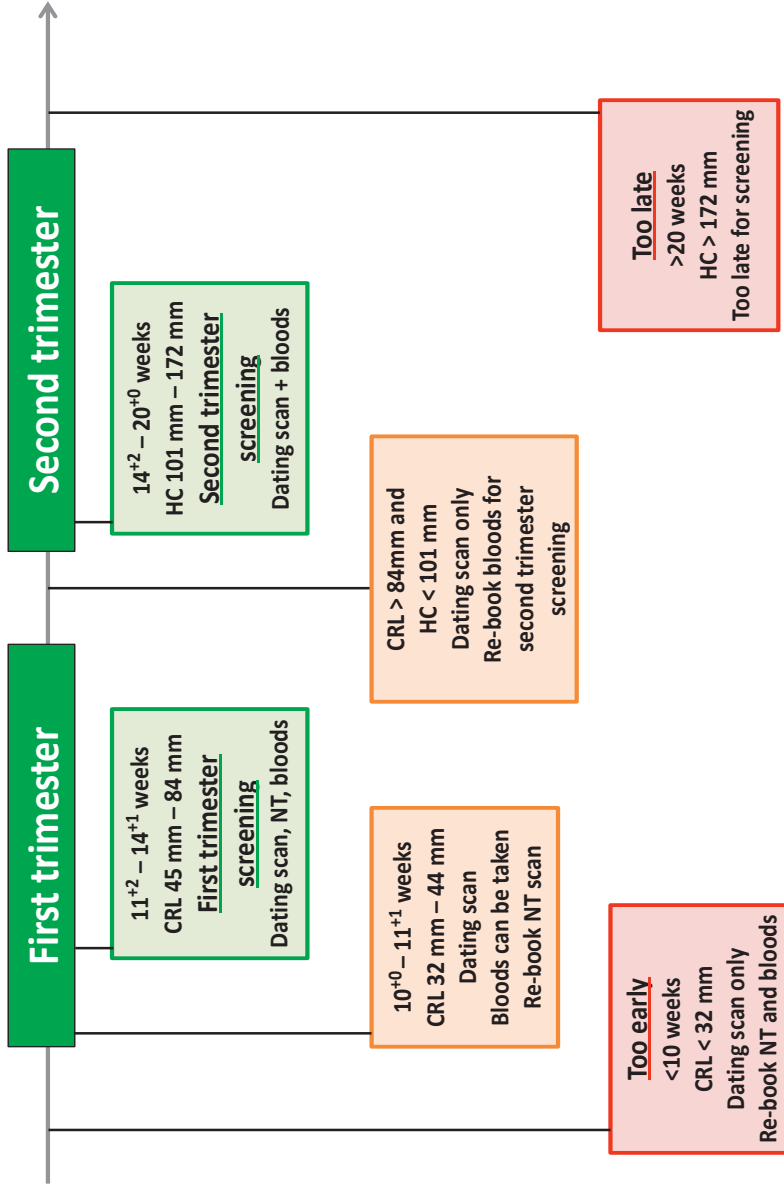
Truncated MoMs

The term truncation is used to describe the process of establishing upper and lower limits to the calculation of MoMs for extreme marker values. This is because extreme levels of marker values are not reliable in the chance calculation. The chance result is therefore calculated using a limited (or truncated) MoM. Where a MoM is truncated this will be shown on the report.

The antenatal screening pathway

Selection of screening tests at different stages of pregnancy

(Use CRL or HC measurements to determine screening pathway rather than gestation in weeks)



The blood sample

- A **minimum of 5 ml** of maternal blood should be taken by venepuncture into a **serum gel blood tube**. Fasting and other patient preparation is not required.
- K⁺-EDTA (haematology) tubes **MUST NOT** be used. If haematology testing is also being carried out, please take the blood sample for this **AFTER** the screening blood sample, as K⁺-EDTA interferes with the analyses.
- The tube **MUST** be labelled with the woman's:
 - First name AND last name
 - Date of birth
 - NHS number/Hospital number (this must also match on the request card)
 - Date/time the sample was taken

Sample storage and transport

- The referring laboratory (if applicable) should send the serum sample/whole blood sample by first class post or by courier to the address given at the start of this handbook packaged according to UN3373 P650 regulations
- Samples should be spun and refrigerated as soon as possible upon receipt in the referring laboratory. Spun serum samples are stable at room temperature for 72 hours and up to 14 days at 4°C.
- Whole blood samples may be sent directly to Bolton laboratory within 48 hours of collection and are stable refrigerated (4°C) for up to 5 days.
- Exposure of samples to temperatures in excess of 30°C can cause rapid deterioration of free beta HCG (within 4 hours) and therefore this should be avoided ¹


Gestation / ultrasound details

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES		Bolton lab. use only		NHS Bolton NHS Foundation Trust	
Please tick which tests you require:			Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!		
T21 <input type="checkbox"/>		Quad test <input type="checkbox"/>		DATE OF ULTRASOUND SCAN	
T13/18 <input type="checkbox"/>		(2 nd trimester)		D D M M Y Y	
HOSPITAL - Name		CONSULTANT		Weeks <input type="text"/> and days <input type="text"/> gestation when scanned	
NHS No.		HOSP No.		CRL <input type="text"/> mm HC <input type="text"/> mm NT <input type="text"/> mm	
<input type="text"/>		<input type="text"/>		EDD D D M M Y Y SONOGRAPHER DQASS CODE <input type="text"/>	
SURNAME:				MATERNAL WEIGHT AT BLOOD SAMPLING <input type="text"/> Kg	
FIRST NAMES:					
ADDRESS:		DOB:		PREGNANCY DETAILS: Y N Y N	
				Current smoker (Within last 2 weeks?) <input type="checkbox"/> <input type="checkbox"/> Type 1 DM on insulin? <input type="checkbox"/> <input type="checkbox"/>	
				Stopped smoking during pregnancy? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM not on insulin? <input type="checkbox"/> <input type="checkbox"/>	
				Type 2 DM on insulin? <input type="checkbox"/> <input type="checkbox"/>	
				Nicotine replacement therapy? <input type="checkbox"/> <input type="checkbox"/> Prev. Down's pregnancy? <input type="checkbox"/> <input type="checkbox"/>	
				Prev. Edwards' pregnancy? <input type="checkbox"/> <input type="checkbox"/>	
				Prev. Patau's pregnancy? <input type="checkbox"/> <input type="checkbox"/>	
FAMILY ORIGIN OF PREGNANT WOMAN:				For IVF Pregnancies only	
<input type="checkbox"/> White		<input type="checkbox"/> Black		Egg origin: Own <input type="checkbox"/>	
<input type="checkbox"/> Other		<input type="checkbox"/> East Asian		Donor <input type="checkbox"/>	
<input type="checkbox"/> South Asian		<input type="checkbox"/> Mixed Black Ethnicity		Extraction date: D D M M Y Y	
		<input type="checkbox"/> Mixed Other Ethnicity		Vanished Twin? <input type="checkbox"/> <input type="checkbox"/>	
				Fetal Pole Seen? <input type="checkbox"/> <input type="checkbox"/>	
				CRL <input type="text"/> mm	
COMMENTS:				Name of requester taking blood	
				Sample date D D M M Y Y Time of sampling	

Key information required to calculate a chance of Down's, Edwards' and Patau's syndrome includes:

- NT measurement (first trimester screening only):**
The measurement is required in mm to one decimal place.
For performance monitoring purposes, the sonographer ID code (e.g. DQASS code) that carried out the NT measurement must be supplied.
- Scan measurements to calculate gestational age**
For first trimester screening, crown rump length (CRL) in mm to one decimal place must be provided ¹. For second trimester screening, a dating scan measurement must be provided. This may be CRL or a head circumference (HC) measurement in mm. The acceptable ranges for HC and CRL measurements are described in the flowchart on page 7.
- Gestational age when scanned**
This complements the scan measurement and allows a cross check/correlation to be carried out in the chance calculation.
- Maternal weight (in kg)** Serum marker concentrations are increased in lower weight women due to reduced blood volume, and decreased in higher weight women due to increased blood volume. The chance calculation corrects for this effect.

Ethnicity

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES		Bolton lab. use only		 NHS Foundation Trust																																					
Please tick which tests you require:			Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!																																						
T21 <input type="checkbox"/> T13/18 <input type="checkbox"/>		Quad test <input type="checkbox"/> (2 nd trimester)		DD DD MM MM YY YY DATE OF ULTRASOUND SCAN																																					
HOSPITAL - Name		CONSULTANT		Weeks <input type="text"/> and days <input type="text"/> gestation when scanned																																					
NHS No. <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>			HOSP No. <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>																																						
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ADDRESS: <input type="text"/>			DOB: <input type="text"/>																																						
PREGNANCY DETAILS: Y N <table border="0" style="width:100%"> <tr> <td>Current smoker (Within last 2 weeks)?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> <td>Type 1 DM on insulin?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> </tr> <tr> <td>Stopped smoking during pregnancy?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> <td>Type 2 DM not on insulin?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> </tr> <tr> <td>Nicotine replacement therapy?/vaping?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> <td>Type 2 DM on insulin?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> </tr> <tr> <td colspan="3">For IVF Pregnancies only</td> <td>Prev. Down's pregnancy?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> </tr> <tr> <td>Egg origin: Own</td> <td><input type="checkbox"/></td> <td></td> <td>Prev. Patau's pregnancy?</td> <td><input type="checkbox"/></td> <td><input type="checkbox"/></td> </tr> <tr> <td>Donor</td> <td><input type="checkbox"/></td> <td></td> <td></td> <td></td> <td></td> </tr> </table>						Current smoker (Within last 2 weeks)?	<input type="checkbox"/>	<input type="checkbox"/>	Type 1 DM on insulin?	<input type="checkbox"/>	<input type="checkbox"/>	Stopped smoking during pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>	Type 2 DM not on insulin?	<input type="checkbox"/>	<input type="checkbox"/>	Nicotine replacement therapy?/vaping?	<input type="checkbox"/>	<input type="checkbox"/>	Type 2 DM on insulin?	<input type="checkbox"/>	<input type="checkbox"/>	For IVF Pregnancies only			Prev. Down's pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>	Egg origin: Own	<input type="checkbox"/>		Prev. Patau's pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>	Donor	<input type="checkbox"/>				
Current smoker (Within last 2 weeks)?	<input type="checkbox"/>	<input type="checkbox"/>	Type 1 DM on insulin?	<input type="checkbox"/>	<input type="checkbox"/>																																				
Stopped smoking during pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>	Type 2 DM not on insulin?	<input type="checkbox"/>	<input type="checkbox"/>																																				
Nicotine replacement therapy?/vaping?	<input type="checkbox"/>	<input type="checkbox"/>	Type 2 DM on insulin?	<input type="checkbox"/>	<input type="checkbox"/>																																				
For IVF Pregnancies only			Prev. Down's pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>																																				
Egg origin: Own	<input type="checkbox"/>		Prev. Patau's pregnancy?	<input type="checkbox"/>	<input type="checkbox"/>																																				
Donor	<input type="checkbox"/>																																								
FAMILY ORIGIN OF PREGNANT WOMAN: <table border="0" style="width:100%"> <tr> <td><input type="checkbox"/> White</td> <td><input type="checkbox"/> Black</td> <td><input type="checkbox"/> Mixed Black Ethnicity</td> </tr> <tr> <td><input type="checkbox"/> Other</td> <td><input type="checkbox"/> East Asian</td> <td><input type="checkbox"/> Mixed Other Ethnicity</td> </tr> <tr> <td><input type="checkbox"/> South Asian</td> <td></td> <td></td> </tr> </table>						<input type="checkbox"/> White	<input type="checkbox"/> Black	<input type="checkbox"/> Mixed Black Ethnicity	<input type="checkbox"/> Other	<input type="checkbox"/> East Asian	<input type="checkbox"/> Mixed Other Ethnicity	<input type="checkbox"/> South Asian																													
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<input type="checkbox"/> Other	<input type="checkbox"/> East Asian	<input type="checkbox"/> Mixed Other Ethnicity																																							
<input type="checkbox"/> South Asian																																									
COMMENTS: <input type="text"/>			Name of requester taking blood Sample date DD DD MM MM YY YY Time of sampling																																						
			CRL <input type="text"/> mm HC <input type="text"/> mm NT <input type="text"/> mm EDD DD DD MM MM YY YY SONOGRAPHER DQASS CODE <input type="text"/> MATERNAL WEIGHT AT BLOOD SAMPLING <input type="text"/> Kg Vanished Twin? <input type="checkbox"/> <input type="checkbox"/> Fetal Pole Seen? <input type="checkbox"/> <input type="checkbox"/> Age of donor at extraction: <input type="text"/> CRL <input type="text"/> mm																																						

- Physiological and genetic variation in serum biochemical marker concentrations occur in different populations, and are taken into account when calculating chance.
- Please refer to the reverse of the request form as a guide to selecting the correct family origin of the women (see page 16).

Pregnancy details

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES		Bolton lab. use only					
Please tick which tests you require:				Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!			
T21 <input type="checkbox"/>		Quad test <input type="checkbox"/>		DATE OF ULTRASOUND SCAN			
T13/18 <input type="checkbox"/>		(2 nd trimester)		D D M M Y Y			
HOSPITAL - Name		CONSULTANT		Weeks <input type="text"/> and days <input type="text"/> gestation when scanned			
NHS No.		HOSP No.		CRL <input type="text"/> mm HC <input type="text"/> mm NT <input type="text"/> mm			
<input type="text"/>		<input type="text"/>		EDD D D M M Y Y SONOGRAPHER DQASS CODE <input type="text"/>			
SURNAME:				MATERNAL WEIGHT AT BLOOD SAMPLING <input type="text"/> Kg			
FIRST NAMES:							
ADDRESS:		DOB:		PREGNANCY DETAILS: Y N Y N			
				Current smoker (Within last 2 weeks)? <input type="checkbox"/> <input type="checkbox"/> Type 1 DM on insulin? <input type="checkbox"/> <input type="checkbox"/>			
				Stopped smoking during pregnancy? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM not on insulin? <input type="checkbox"/> <input type="checkbox"/>			
				Type 2 DM on insulin? <input type="checkbox"/> <input type="checkbox"/>			
				Nicotine replacement therapy?/vaping? <input type="checkbox"/> <input type="checkbox"/> Prev. Down's pregnancy? <input type="checkbox"/> <input type="checkbox"/>			
				Prev. Edwards' pregnancy? <input type="checkbox"/> <input type="checkbox"/>			
				Prev. Patau's pregnancy? <input type="checkbox"/> <input type="checkbox"/>			
FAMILY ORIGIN OF PREGNANT WOMAN:				<u>For IVF Pregnancies only</u>			
<input type="checkbox"/> White		<input type="checkbox"/> Black		Egg origin: Own <input type="checkbox"/>			
<input type="checkbox"/> Other		<input type="checkbox"/> East Asian		Donor <input type="checkbox"/>			
<input type="checkbox"/> South Asian		<input type="checkbox"/> Mixed Black Ethnicity		Extraction date: D D M M Y Y			
		<input type="checkbox"/> Mixed Other Ethnicity		Age of donor at extraction: <input type="text"/>			
				Vanished Twin? Y N <input type="checkbox"/> <input type="checkbox"/>			
				Fetal Pole Seen? <input type="checkbox"/> <input type="checkbox"/>			
				CRL <input type="text"/> mm			
COMMENTS:				Name of requester taking blood			
				Sample date D D M M Y Y Time of sampling			

Further critical information required for reliable chance calculation includes:

- **Smoking status**

Some serum marker concentrations are affected by smoking and this is corrected for in the risk calculation. The effect is less for nicotine replacement which is not currently corrected for but may be in the future.

- **Diabetic status**

Serum marker concentrations are affected in Type 1 and Type 2 diabetics and in women on insulin. This is taken into account in the risk calculation.

- **Previous Down's/Edwards'/Patau's syndrome pregnancy**

Women with a previous trisomy pregnancy are at higher chance of subsequent affected pregnancies and this is accounted for in the chance calculation.

IVF Pregnancy

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES		Bolton lab. use only			
Please tick which tests you require:			Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!		
T21 <input type="checkbox"/> T13/18 <input type="checkbox"/>		Quad test <input type="checkbox"/> (2 nd trimester)		DD DD MM MM YY YY DATE OF ULTRASOUND SCAN	
HOSPITAL - Name		CONSULTANT		Weeks <input type="text"/> and days <input type="text"/> gestation when scanned	
NHS No. <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>		HOSP No. <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>		CRL <input type="text"/> mm HC <input type="text"/> mm NT <input type="text"/> mm	
SURNAME:			EDD DD DD MM MM YY YY SONOGRAPHER DQASS CODE <input type="text"/>		
FIRST NAMES:			MATERNAL WEIGHT AT BLOOD SAMPLING <input type="text"/> Kg		
ADDRESS:		DOB:	PREGNANCY DETAILS: Y N		
			Current smoker (Within last 2 weeks)? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Stopped smoking during pregnancy? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Nicotine replacement therapy?/vaping? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Type 1 DM on insulin? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Type 2 DM not on insulin? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Type 2 DM on insulin? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Prev. Down's pregnancy? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Prev. Edwards' pregnancy? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Prev. Patau's pregnancy? <input type="checkbox"/> Y <input type="checkbox"/> N		
FAMILY ORIGIN OF PREGNANT WOMAN:			For IVF Pregnancies only		
<input type="checkbox"/> White		<input type="checkbox"/> Black	<input type="checkbox"/> Mixed Black Ethnicity		Egg origin: Own <input type="checkbox"/>
<input type="checkbox"/> Other		<input type="checkbox"/> East Asian	<input type="checkbox"/> Mixed Other Ethnicity		Donor <input type="checkbox"/>
<input type="checkbox"/> South Asian					Extraction date: DD DD MM MM YY YY
COMMENTS:			Age of donor at extraction: <input type="text"/>		
			Vanished Twin? <input type="checkbox"/> Y <input type="checkbox"/> N		
			Fetal Pole Seen? <input type="checkbox"/> Y <input type="checkbox"/> N		
			CRL <input type="text"/> mm		
			Name of requester taking blood		
			Sample date DD DD MM MM YY YY Time of sampling		

The laboratory is able to process samples from IVF pregnancies. The age of the egg at the time of extraction is important for accurate Down's, Edward's and Patau's syndrome chance calculation, hence the request form should include:

The origin of the eggs used; whether they are the woman's 'own' or 'donor' eggs.

If from a donor the age of the donor at extraction is required.

If the eggs are from a donor, the donor's age will be used in the chance calculation. If the donor age is not known then a donor age of 35 will be inputted as the age¹

Vanished twin

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES			Bolton lab. use only		 Bolton NHS Foundation Trust	
Please tick which tests you require:			Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!			
T21 <input type="checkbox"/>		Quad test <input type="checkbox"/>		DATE OF ULTRASOUND SCAN [D][D][M][M][Y][Y]		
T13/18 <input type="checkbox"/>		(2 nd trimester)		Weeks [] and days [] gestation when scanned		
HOSPITAL - Name		CONSULTANT		CRL [] mm HC [] mm NT [] mm		
NHS No.		HOSP No.		EDD [D][D][M][M][Y][Y] SONOGRAPHER DQASS CODE []		
SURNAME:				MATERNAL WEIGHT AT BLOOD SAMPLING [] Kg		
FIRST NAMES:				PREGNANCY DETAILS: Y N Y N Current smoker (Within last 2 weeks)? <input type="checkbox"/> <input type="checkbox"/> Type 1 DM on insulin? <input type="checkbox"/> <input type="checkbox"/> Stopped smoking during pregnancy? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM not on insulin? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM on insulin? <input type="checkbox"/> <input type="checkbox"/> Nicotine replacement therapy?/vaping? <input type="checkbox"/> <input type="checkbox"/> Prev. Down's pregnancy? <input type="checkbox"/> <input type="checkbox"/> Prev. Edwards' pregnancy? <input type="checkbox"/> <input type="checkbox"/> Prev. Patau's pregnancy? <input type="checkbox"/> <input type="checkbox"/>		
ADDRESS:		DOB:		For IVF Pregnancies only Egg origin: Own <input type="checkbox"/> Donor <input type="checkbox"/> Extraction date: [D][D][M][M][Y][Y] Age of donor at extraction: []		
FAMILY ORIGIN OF PREGNANT WOMAN: <input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Mixed Black Ethnicity <input type="checkbox"/> Other <input type="checkbox"/> East Asian <input type="checkbox"/> Mixed Other Ethnicity <input type="checkbox"/> South Asian			Vanished Twin? <input type="checkbox"/> <input type="checkbox"/> Y N Fetal Pole Seen? <input type="checkbox"/> <input type="checkbox"/> CRL [] mm			
COMMENTS:			Name of requester taking blood Sample date [D][D][M][M][Y][Y] Time of sampling			

The presence of a vanished twin can impact the screening markers in the blood. If a vanished twin is seen in the first trimester screening can be performed if there is no visible fetal pole. If a fetal pole is seen then the chances can be calculated using age and NT measurements only. In the second trimester the quadruple test can be performed in the case of a vanished twin but it will not perform as well as in singleton pregnancies. Changes to the way the vanished twin risks are calculated are to be introduced in 2024/2025 by FASP.

Information on the back of the request form

FAMILY ORIGIN OF PREGNANT WOMAN:

WHITE	England, Scotland, Wales, Northern Ireland Austria, Belgium, Ireland, France, Germany, Netherlands, Scandinavia, Switzerland etc. Any other European family origins eg. Australia, USA, South Africa Cyprus, Greece, Turkey, Italy, Portugal, Spain, Sardinia Any other Mediterranean country Albania, Czechia, Poland, Romania, Russia etc
OTHER	North Africa, South America, Middle East
BLACK	Caribbean Islands, Africa (excluding North Africa)
SOUTH ASIAN	India or African- Indian, Pakistan, Bangladesh, Sri Lanka
EAST ASIAN	China, Thailand, Malaysia, Vietnam, Philipines, Cambodia, Indonesia, Burma etc
MIXED BLACK ETHNICITY	Any mixed ethnicity including Black ethnicity
MIXED OTHER ETHNICITY	Any mixed ethnicity not including Black ethnicity

IVF PREGNANCY

If donor eggs are used, the age of the eggs at the time of extraction should be supplied.

(If the eggs are from a donor it is the eggs' age that will be used to calculate the chances of Down's, Edwards' or Patau's syndrome)

Tel: (01204) 390424

R21c
Version 9

Twin pregnancies

In the first trimester antenatal screening for Down's, Edwards' and Patau's syndromes may be performed in twin pregnancies¹. Only screening for Down's syndrome can be performed in the second trimester. A specific twin pregnancy antenatal screening request form must be used (see below). The blood sample should be taken under the same conditions as in a singleton pregnancy.

ANTENATAL SCREENING SERVICE FOR DOWN'S (T21), EDWARDS' (T18)/PATAU'S (T13) SYNDROMES FOR TWINS		Bolton lab. use only	
Please tick which tests you require:		Min 5ml PLAIN BLOOD REQUIRED - NO ANTICOAGULANT!	
T21 <input type="checkbox"/> T13/18 <input type="checkbox"/>	Quad test <input type="checkbox"/> (2 nd trimester)	DATE OF ULTRASOUND SCAN <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>	
HOSPITAL - Code or Name	CONSULTANT	CRL Fetus 1 <input type="text"/> mm	HC Fetus 2 <input type="text"/> mm
NHS No. <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>	HOSP No.	NT Fetus 1 <input type="text"/> mm	NT Fetus 2 <input type="text"/> mm
SURNAME:		EDC <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>	SONOGRAPHER ID CODE <input type="text"/>
FIRST NAMES:		Monochorionic? <input type="checkbox"/> Dichorionic? <input type="checkbox"/> Weight <input type="text"/> kg	
ADDRESS:	DOB:	PREGNANCY DETAILS: Y N Y N Current smoker (Within last 2 weeks)? <input type="checkbox"/> <input type="checkbox"/> Type 1 DM on insulin? <input type="checkbox"/> <input type="checkbox"/> Stopped smoking during pregnancy? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM not on insulin? <input type="checkbox"/> <input type="checkbox"/> Nicotine replacement therapy? <input type="checkbox"/> <input type="checkbox"/> Type 2 DM on insulin? <input type="checkbox"/> <input type="checkbox"/> For IVF Pregnancies only Egg origin: Own <input type="checkbox"/> Prev. Down's pregnancy? <input type="checkbox"/> <input type="checkbox"/> Donor <input type="checkbox"/> Prev. Edwards' pregnancy? <input type="checkbox"/> <input type="checkbox"/> Prev. Patau's pregnancy? <input type="checkbox"/> <input type="checkbox"/> Extraction date: <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> Age of donor extraction: <input type="text"/>	
FAMILY ORIGIN OF PREGNANT WOMAN: <input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Mixed Black Ethnicity <input type="checkbox"/> Other <input type="checkbox"/> East Asian <input type="checkbox"/> Mixed Other Ethnicity <input type="checkbox"/> South Asian		NAME OF REQUESTER TAKING BLOOD:..... SIGNATURE:..... Sample Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>	
COMMENTS:			

First trimester

In both mono- and dichorionic pregnancies, NT and CRL measurements are required for both fetuses¹.

- In dichorionic twin pregnancies 2 chances will be issued, one per fetus per condition screened. (i.e.T21 and/or T13/18)
- In monochorionic pregnancies one chance will be issued per condition screened and a mean of the NT measurements used in the calculation. This is because monochorionic twins are almost always genetically identical.

For further information regarding multiple pregnancies please contact the laboratory.

Second trimester

In mono- and dichorionic pregnancies, CRL and/or HC measurements are required for both fetuses¹. A single Down's syndrome pregnancy chance is reported.

Vanished twin and bleeding

Vaginal bleeding is not thought to affect the chance calculation for Down's syndrome, Edwards' Syndrome and Patau's syndrome.

If an empty second pregnancy sac is seen on a first trimester NT scan, screening can continue as normal. If a second pregnancy sac contains a non-viable fetus or fetal pole, then the markers may be affected and combined first trimester screening should not be undertaken. The laboratory can calculate the chance using NT and maternal age alone in this case if required. In the second trimester the quadruple test can be undertaken in the case of a vanished twin.¹ FASP Guidance on vanished twins is expected to change in 2025.

Chance interpretation

Both the first trimester combined test for Down's and Edwards' and Patau's syndromes and the second trimester quad test for Down's syndrome categorise women into two groups:

1. Higher chance of having an affected pregnancy
2. Lower chance of having an affected pregnancy

Higher chance

In both the first trimester test and the second trimester test, a woman is reported as being at higher chance of having an affected pregnancy when the chance is **1 in 150** or higher¹. The highest chance that will be reported is **higher than 1 in 2** (displayed as 1 in 2)

Lower chance

In both the first trimester test and the second trimester test, a woman is reported as being lower chance of having an affected pregnancy when the chance is lower than **1 in 150**¹.

A lower chance result does not exclude the possibility of an affected pregnancy.


The lowest chance that will be reported is **less than 1 in 5000** (displayed as 1 in 5000).

Reporting of results

The laboratory offers a three working day turn-around time from receipt of the blood sample to reporting of results. All higher-chance results are telephoned or emailed directly to the user, and electronic or paper reports are issued for all higher and lower chance reports.

Ante-Natal Screening Laboratory

PATIENT REPORT



NHS Number:

CHI Number:

Name: TEST TEST

DOB: 12/10/2004

Hospital Number:

Ethnicity: Caucasian

Address: 12 TEST ROAD
ANYTIME
BOLTON
BL4 0JR

Post Code: BL4 0JR

REQUESTOR

Address: **Royal Bolton Hospital**
Minerva Road
Farnworth
Greater Manchester
BL4 0JR

PREGNANCY DETAILS

Mat Age at EDD: 20 years 4 months

Smoking: None

Fetuses: 1

EDD: 01/03/2025

SPECIMEN DETAILS

Barcode: 24T128506H

Maternal Weight: 76 kg

Gestational Age at Collection Date: 12 Weeks 5 Days

Scan Date: 22/08/2024

Sample Collected: 22/08/2024

Date Received: 22/08/2024

Gestation at Scan date: 12 Weeks 5 days

CRL: 63 mm

HC:

Specimen tests:

Test	Concentration	Unit	Median	MoM	Corr. MoM
hCGb	37	U/L	38.18	0.97	1.04
NT	1.8	mm			
PAPP-A	750	mIU/L	2498.45	0.3	0.32

CHANCES

Down's syndrome

Chance rating: LOWER

Age chance: 1 in 1500

Down's chance at term: 1 in 1300

Cutoff: 1 in 150

Combined Edwards' and Patau's

Chance rating: LOWER

Age chance: 1 in 5000

T18/T13 chance at term: 1 in 6000

Cutoff: 1 in 150

Tel 01204 390424
Please note: This interpretation assumes that patient and specimen details are accurate
UKAS Accredited

1/2

22/08/2024

To enable reporting of results electronically, please contact the laboratory. Lower chance patient letters are also available to download

Monitoring performance

The screening test is expected to detect 85% of T21 pregnancies and 80% of T13/18 pregnancies in the first trimester and 80% of T21 pregnancies in the second trimester with approximately 3% of women having a higher chance result.

This data is collected and monitored nationally by NCARDRS.

Measures of performance of the antenatal screening programme include:

1. The detection rate and screen positive rate by DQASS
2. Achieving a three day laboratory turnaround for 98.5% of screening reports²
3. Performance on external quality assurance schemes (see below)
4. National Screening Committee key performance indicators (e.g.FA4- Unacceptable samples rate).²

Quality

All screening strategies are subject to external quality assessment, audit, and assurance schemes. This includes participation in the Down's Quality Assurance Support Service (DQASS). Every six months non-identifiable biochemistry and ultrasound NT data are sent to DQASS where the median values and performance of the screening test are assessed against the recommended programme outcomes. DQASS issues a full report, along with a summary report to the Chief Executive of all participating Trusts, as well as all relevant Public Health England departments. It is essential that all labs are UKAS Accredited and participate in National External Quality Assurance Schemes (UK NEQAS). The department at Bolton fully complies with these requirements.

Interference

Very occasionally the laboratory is unable to provide a reliable screening result due to interference in the immunoassays used to measure the hormones in the blood. The laboratory will contact the screening midwife with further advice if assay interference is suspected in a sample.

The team

Carolyn Williams

Consultant Clinical Biochemist & Director of Antenatal Screening

James Osborne

Principal Clinical Biochemist

Karina Hambridge

Antenatal Screening Laboratory Manager

Trine Nielsen

Antenatal Screening Laboratory Section Manager

Cassie Hesketh, Lewis Roddie, Jenny Gorton

Healthcare Science Practitioners

References

1. Screening for Down's Edwards' and Patau's syndrome (FASP Handbook) [Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome - GOV.UK \(www.gov.uk\)](http://www.gov.uk)
2. Fetal anomaly screening standards 2022. [Fetal anomaly screening standards valid for data collected from 1 April 2022 - GOV.UK \(www.gov.uk\)](http://www.gov.uk)

Abbreviations

AFP	Alpha-feto protein
βhCG	Free-beta human chorionic gonadotrophin
CRL	Crown rump length
DQASS	Down's syndrome Quality Assurance Support Service
HC	Head circumference
IVF	In-vitro fertilisation
MoM	Multiples of the median
NCARDS	National Congenital Anomaly register Fetal anomaly screening standards valid for data collected from 1 April 2022 - GOV.UK (www.gov.uk)
NEQAS	National External Quality Assessment Scheme
NSC	National Screening Committee
NT	Nuchal translucency
PAPPA	Pregnancy associated plasma protein A
uE3	Unconjugated oestriol
UKAS	United Kingdom Accreditation Service

Summer 2024
Version 5