

## APPENDIX 1

## WOMEN'S VIEWS OF DIAGNOSTIC TESTS FOR DOWN SYNDROME DURING PREGNANCY

## BACKGROUND

Diagnostic tests for Down syndrome can vary in a number of ways, including:

- **Accuracy:** Tests can vary in how accurate they are. The tests described here can identify 95%, 99% or 100% of babies with Down syndrome during pregnancy. (*Please see the separate sheet for more information on accuracy.*)
- **Time in pregnancy when results are received:** Tests vary in when they can be done and how long it takes to get a result. With the tests described here you could get your results at 10 weeks, 12 weeks or 16 weeks in pregnancy.
- **Risk of miscarriage:** Some tests have no risk of miscarriage. Others have a small risk (1% risk) of miscarriage. A 1% risk means that one out of every 100 women who have a test will have a miscarriage.
- **Information from tests:** Generally, prenatal tests can give two types of information.

**Simple information:** These tests will only tell you whether or not the baby has one of the three most common conditions caused by changes in the chromosomes – Down syndrome, Edwards syndrome or Patau syndrome.

**Comprehensive information:** These tests will tell you whether or not the baby has one of the three most common conditions caused by changes in the chromosomes (including Down syndrome) and will also give information about whether some rare conditions are present, which may cause learning disability, developmental delay or other health problems.

Tests that give comprehensive information look in detail at all of the baby's chromosomes. Changes can include additional, missing or rearranged chromosome material. The effects of these changes can vary and severity will depend upon the parts of chromosomes that are involved. Some changes can result in severe disabilities, but these are very rare. Furthermore, in some cases, it may be difficult to predict what problems, if any, the child may have and others may only become significant in adult life.

## SECTION A: THE FOLLOWING QUESTIONS ASK WHAT FEATURES OF PRENATAL TESTS ARE MOST IMPORTANT TO YOU.

1. Please rank the following test features in order of importance to you.

(1: most important; 6: least important)

- Test as early as possible.
- Test with the greatest level of accuracy.
- Test that has a low financial cost for patients.
- Test as safe as possible, with the lowest possible risk of a miscarriage.
- Test that diagnoses Down syndrome and gives additional information about conditions that may cause learning disability, developmental delay or other health problems, even if there is a possibility of finding results of unknown significance.
- Test with the shortest waiting time for results.

2. Please read the following and answer the question according to your own views.

Candice is 10 weeks pregnant and when she has her first antenatal appointment she is told about two different diagnostic tests for Down syndrome. The doctor tells her that she does not have to take either test if she does not want to. It is her choice.

**Test 1:** The test is 99% accurate and will tell you whether or not the baby has Down syndrome, Edwards syndrome or Patau syndrome. This test will not give any additional information about the health of the baby. The test is a blood test and there is no risk of miscarriage. However, if the result is positive, your doctor may recommend an invasive test, which has a 1% risk of miscarriage.

**Test 2:** The test is 100% accurate and will tell you whether or not the baby has Down syndrome, Edwards syndrome or Patau syndrome. This test will also give additional information about rare conditions that may cause learning disability, developmental delay or other health problems. This is an invasive test and there is a 1% risk of miscarriage.

Imagine you are Candice and tell us what you would choose to do.

- I would have Test 1.
- I would have Test 2.
- I do not wish to have either test.

**SECTION B: THE FOLLOWING QUESTIONS REQUIRE YOU TO COMPARE BETWEEN TWO PRENATAL TESTS FOR DOWN SYNDROME AND INDICATE WHICH ONE YOU WOULD CHOOSE.**

There are ten choices listed below. Each choice involves two different diagnostic tests that will tell you during pregnancy whether or not the baby has Down syndrome. The tests are hypothetical and are not available in any hospital.

Please consider each choice separately and tick the box to show which option you would prefer: A, B or neither.

Choice 1	Test A	Test B
Accuracy	99%	100%
Time of results	10 weeks	12 weeks
Risk of miscarriage	Small risk (1%)	No risk
Information from test	Comprehensive information	Simple information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 2	Test A	Test B
Accuracy	100%	95%
Time of results	10 weeks	12 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 3	Test A	Test B
Accuracy	100%	95%
Time of results	16 weeks	10 weeks
Risk of miscarriage	Small risk (1%)	No risk
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 4	Test A	Test B
Accuracy	95%	99%
Time of results	10 weeks	12 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 5	Test A	Test B
Accuracy	95%	100%
Time of results	16 weeks	10 weeks
Risk of miscarriage	Small risk (1%)	No risk
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 6	Test A	Test B
Accuracy	95%	99%
Time of results	12 weeks	16 weeks
Risk of miscarriage	Small risk (1%)	No risk
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 7	Test A	Test B
Accuracy	99%	100%
Time of results	12 weeks	16 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 8	Test A	Test B
Accuracy	100%	95%
Time of results	12 weeks	16 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Comprehensive information	Simple information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 9	Test A	Test B
Accuracy	99%	100%
Time of results	16 weeks	10 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Simple information	Comprehensive information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

Choice 10	Test A	Test B
Accuracy	95%	99%
Time of results	16 weeks	10 weeks
Risk of miscarriage	No risk	Small risk (1%)
Information from test	Comprehensive information	Simple information

Which test would you prefer? (Choose only one)

Test A  Test B  Neither

**SECTION C: SOME QUESTIONS ABOUT YOU.**

1. How old are you? \_\_\_\_\_
2. What is your relationship status?
  - Single  In a relationship
3. What is your highest educational qualification? (*Please choose one*)
  - No qualification
  - GCSE or GCE 'O' level
  - GCE 'A' level or similar
  - Higher education or similar
  - Other (please specify): \_\_\_\_\_
4. Do you have a religious faith?
  - Yes  No
 If yes, please specify: \_\_\_\_\_
5. Which of the following best describes you? (*Please choose one*)
  - Chinese  Japanese  Indian
  - Malay  Caucasian  Pakistani
  - Black – African  Black – Other  Bangladeshi
  - Others
6. Do you have any children?
  - Yes, I have \_\_ child(ren)  No
7. Are you currently pregnant?
  - Yes  No
 If yes, how many weeks pregnant are you? \_\_\_\_\_
8. Have you had a previous miscarriage?
  - Yes  No
9. I have had or am likely to have prenatal testing for Down syndrome during pregnancy.
  - Strongly agree  Agree  Disagree  Strongly disagree
 If you chose strongly agree or agree, please indicate which statement best matches why you have chosen to have a prenatal test.
  - So I can plan and prepare for the possibility of having a child with Down syndrome.
  - To help me make a decision around termination of pregnancy.
  - I would want as much information about the baby as possible.
  - Because my partner or family would want me to.
  - Because my doctor suggested it.
  - Because it is part of the routine prenatal care that women have in pregnancy.
  - Other (please specify) \_\_\_\_\_
 If you chose strongly disagree or disagree, please indicate which statement best matches why you have chosen not to have a prenatal test.
  - I would never have a termination of pregnancy, so there is no point in having a test.
  - It would cause a lot of anxiety if I found out the baby had a health problem.
  - I would not want to know and then have to make a decision about what to do next.
  - Because my partner or family would not want me to have a test.
  - I would prefer not to know.
  - Other (please specify) \_\_\_\_\_
10. Do you have a child with Down syndrome?
  - Yes  No
11. Does anyone you know have a child with Down syndrome?
  - Yes  No

## APPENDIX 2

**Table I. Ranked preferences for prenatal test attributes by women.**

Attribute	%					
	1	2	3	4	5	6
Early test	18.1	15.6	17.7	27.9	12.7	8.0
Accuracy	27.9	32.1	26.6	9.3	3.0	1.3
Cost	5.1	9.7	8.0	18.1	29.5	29.5
Safety	41.3	22.4	20.7	11.0	4.2	0.4
Full information	10.6	18.1	22.8	19.8	19.4	9.3
Time of results	1.3	4.6	4.2	10.6	30.0	49.4

Some women did not rank all six attributes. 1: most important; 6: least important

**Table II. Ranked preferences for prenatal test attributes by health professionals.**

Attribute	%					
	1	2	3	4	5	6
Early test	6.3	15.6	20.3	21.9	15.6	20.3
Accuracy	64.1	25.0	6.3	3.1	1.6	0
Cost	6.3	4.7	18.8	18.8	28.1	23.4
Safety	18.8	46.9	20.3	9.4	4.7	0
Full information	4.7	6.3	28.1	23.4	7.8	29.7
Time of results	0	3.1	6.3	23.4	42.2	25.0

Five health professionals did not complete this section. 1: most important; 6: least important.

**Table III. Results of conditional logit regression analysis for women and HPs.**

Attribute	Women (n = 301)		HPs (n = 69)		p-value
	Coefficient (95% CI)*	p-value	Coefficient (95% CI)*	p-value	
Accuracy	0.195 (0.167–0.222)	< 0.0001	0.322 (0.259–0.386)	< 0.0001	0.0003
Time of results	-0.075 (-0.095 to -0.055)	< 0.0001	-0.131 (-0.173 to -0.089)	< 0.0001	0.0180
Full information	0.401 (0.305–0.496)	< 0.0001	0.485 (0.278–0.691)	< 0.0001	0.4696
No miscarriage risk	1.250 (1.149–1.350)	< 0.0001	0.802 (0.582–1.023)	< 0.0001	0.0003

\*No. of observations = 8,982; pseudo-R<sup>2</sup> = 0.2969. \*No. of observations = 2,052; pseudo-R<sup>2</sup> = 0.3876. CI: confidence interval; HPs: health professionals

**Table IV. Women and health professionals (HPs) who did not make trade-offs with their choices.**

Parameter	No. (%)	
	Women (n = 301)	HPs (n = 69)
Chose tests with the highest accuracy for all options	19 (6.3)	10 (14.5)
Chose tests with the earliest time for all options	1 (0.3)	1 (1.4)
Chose tests with full information for all options	5 (1.7)	3 (4.3)
Chose tests with no risk for all options	87 (28.9)	6 (8.7)
Chose neither for all options	11 (3.7)	0 (0)
Total	123 (40.9)	20 (29.0)

Table V. Percentage of women, according to demographic categories, who would select NIPT, IPD or neither.

Parameter	No.	No. (%)		
		NIPT (n = 133)	IPD (n = 117)	Neither (n = 36)
<b>Age (yr)</b>				
< 35	214	94 (43.9)*	93 (43.5)	27 (12.6)
≥ 35	61	38 (62.3)	18 (29.5)	5 (8.2)
<b>Has religious faith</b>				
No	72	35 (48.6)	35 (48.6)	2 (2.8)
Yes	200	89 (44.5)	78 (39.0)	33 (16.5)
<b>Declared faith</b>				
Buddhist	28	14 (50.0)	12 (42.9)	2 (7.1)
Christian	53	33 (62.3)*	9 (17.0)	11 (20.8)
Hindu	26	13 (50.0)	13 (50.0)	0 (0.0)
Muslim	55	21 (38.2)	29 (52.7)	5 (9.1)
Sikh	3	1 (33.3)	2 (66.7)	0 (0.0)
Taoist	3	2 (66.7)	1 (33.3)	0 (0.0)
Other	4	1 (25.0)	2 (50.0)	1 (25.0)
<b>Already has children</b>				
No	158	74 (46.8)	63 (39.9)	21 (13.3)
Yes	121	53 (43.8)	53 (43.8)	15 (12.4)
<b>Previous miscarriage</b>				
No	213	91 (42.7)	95 (44.6)	27 (12.7)
Yes	57	33 (57.9)*	15 (26.3)	9 (15.8)
<b>Knows a child with DS</b>				
No	216	95 (44.0)	98 (45.4)	23 (10.6)
Yes	50	26 (52.0)†	12 (24.0)	12 (24.0)
<b>Healthcare type</b>				
Private	183	97 (53.0)*	64 (35.0)	22 (12.0)
Subsidised	101	35 (34.7)†	52 (51.5)	14 (13.9)
<b>Ethnicity</b>				
Chinese	100	61 (61.0)*	31 (31.0)	8 (8.0)
Indian	52	18 (34.6)†	30 (57.7)	4 (7.7)
Malay	48	20 (41.7)	20 (41.7)	8 (16.7)
Others	67	23 (34.3)	28 (41.8)	16 (23.9)

A binomial test was used to determine whether there was a significant difference between the number of women choosing IPD or NIPT in each category. \*p < 0.001 was statistically significant. †p < 0.05 was statistically significant. DS: Down syndrome; IPD: invasive prenatal diagnosis; NIPT: noninvasive prenatal testing