

Antenatal testing

Introduction

This activity will introduce students to some of the issues involved in medical screening during pregnancy using different techniques.

Students will also think about false positive and false negative results in relation to such tests, and the possible consequences for prospective parents and society.

The activity

Students are given a short clip from a newspaper report, and asked a number of questions about what they have read.

Once they have finished, you might ask one or two students to present their answer to the final question. It may also be useful to ask students to try to explicitly identify which of the HSW ideas they have used in answering the questions.

Figure 1: risk of Down's syndrome by maternal age Cuckle HS et al (1987) BrJObstGynae **94**, 387-402

Figure 2: Data taken from

<http://www.wolfson.qmul.ac.uk/ndscr/reports/NDCSRreport05.pdf>

Suggested answers to questions

1. A mother is told that "she has a 1 in 250 risk of her baby having Down's syndrome". What does this statement mean?
For every 250 babies born to mothers with the same screening results 1 will have Down's syndrome, 0.4% risk of having a baby with Down's..
2. In such a screening programme, if 100 unborn babies have Down's syndrome, how many of them will be undetected?
25 babies undetected
3. This is a false negative result. What are the possible consequences of such a result?
Parents go through pregnancy assuming everything is normal.
 - *May experience shock and worry after the birth*
 - *May find it hard to bond with the baby*
4. Briefly describe how these two tests are carried out.
Chorionic villus - a fine needle, guided by ultrasound, is passed through the woman's abdomen, or vagina and cervix, into the developing placenta and a tiny piece is removed. A chromosome analysis of these cells is then carried out.

Resources

Textbook p. 87-90

How Science Works

Aa We can never be sure that an observation is accurate or that a measurement tells us the 'true' value of the quantity we are measuring. We may be influenced by what we expect. Random and/or systematic errors can arise from limitations of the measuring equipment used, or our skill in using it. A single measurement of anything is therefore inherently risky.

Ag Data can be presented and analysed in a variety of formats including graphs with linear scales.

Gf To make an informed decision about the management of a given risk, we need to take account both of the probability of the event occurring, and the seriousness of the consequences if it did. This is particularly difficult in the case of events of very low probability, but with very serious consequences if they occur.

Hb Decision makers aim to make evidence-based decisions, taking into account factors that include:

technical feasibility, benefits expected, economic cost, risks to human health and well-being, risks to the environment.

Hi Decisions and opinions on ethical issues are based on values. An individual's view on an issue may be based upon a religious or moral position.

Hj Some decisions involve balancing the rights of certain individuals and groups against those of others.

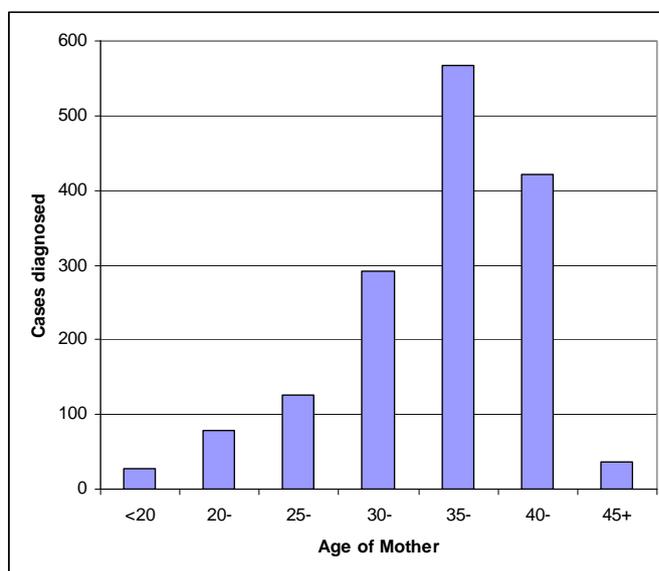
Hk A utilitarian approach is to argue that the right decision or choice is the one that leads to the greatest good for the largest number. It can also be argued, however, that some actions are wrong, even if they lead to good outcomes.

Amniocentesis - A fine needle, guided by ultrasound, is inserted into the amniotic fluid. This fluid contains foetal cells that are cultured in the laboratory and then the chromosomes analysed in detail for abnormalities.

5. Why is it important to have a low percentage of false positives in the initial screening programme?
 - *minimise anxiety*
 - *the additional tests (amniocentesis and cvs) both carry risk of increased miscarriage so could end up with more miscarriages.*
6. Suggest why amniocentesis and chorionic villus sampling are not routinely offered to younger women.

additional risk of miscarriage greater than the risk of Down's syndrome.
7. Some women refuse either the screening tests or the offer of further testing. Suggest 2 reasons for this.
 - *May not wish the increased the risk of miscarriage.*
 - *May have ethical or religious objections to abortion.*

8



9. Describe the trends in the data for 2005 between maternal age and diagnosis of Down's Syndrome.
 - *Increasing number of cases of Down's syndrome diagnosed as age of mother increases - as suggested by table of risk .*
 - *Big decrease in number of cases above age of 45 - fewer women have babies at this age*
 - *Small number of cases below age of 20 - as suggested by table of risk.*
10. Write a letter to the Health secretary explaining in detail your view on compulsory initial screening for Down's Syndrome.

Student answers should include HSW ideas such as:

- **Aa** - limitations of data - false positive and negative results
- **Gf** - Management of risk - probability of risk and consequence of the risk
- **Hb**- evidence based decisions based on benefits expected, economic cost
- **Hi** - ethical and religious values
- **Hj** - balancing rights of individuals/groups against those of others
- **Hk** - utilitarian or absolute values

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Introduction

Read the newspaper extract given below, and then answer the questions that follow.

Mum campaigns for more tests.

Knowing that their baby is healthy is what every parent wants to hear. So when Mary Kilbride (30) became pregnant with her first child she, and her husband Alec (35), agreed that they would have the two recommended ultrasound scans at around 12 and 20 weeks.

Both scans - taken at their local NHS trust - showed that everything appeared to be normal. Yet their son Wilf was born with Down's syndrome.

This was such a shock that Mary found it difficult to bond with her new son for the first few weeks. "It was terrible to start with," says Alec, "we weren't expecting any problems, so when Wilf was born, Mary couldn't cope. She blamed the hospital for his birth, and felt that she should have been warned.

Whilst we wouldn't have considered an abortion," Alec added, "we needed time to adjust. Luckily, we've come to terms with how Wilf is, and couldn't ask for a happier or more loving little boy."

"We could have had more tests," says Mary, "Tests which would have given us a much clearer answer, but I wasn't offered them because I was too young. I think that is wrong, and all mothers at risk should at least be told about the extra tests."

There are a number of medical tests which can be used to screen for Down's syndrome including a maternal blood screening test at 14 weeks and an ultrasound scan between 10-13 weeks. Many hospitals in the UK offer a combination of initial screening tests for Down's to pregnant women.

These are combined with the mother's age to give a value for the likely risk of having a child with Down's syndrome.

1. Following screening a mother is told that "she has a 1 in 250 risk of her baby having Down's syndrome". What does this statement mean?

2. The government wants the NHS to implement a screening programme which has detection rate of at least 75% with a false positive rate of 3% or less.

In such a screening programme, if 100 unborn babies have Down's syndrome, how many of them will be undetected?

3. The extract describes a false negative result. What are the possible consequences of such a result?
4. Once the initial screening has been carried out, women who have been identified as having a high risk of having a Down's syndrome baby will be offered further *diagnostic* tests which can, in most cases, provide a more accurate diagnosis.

These tests are chorionic villus sampling (CVS) and amniocentesis

Briefly describe how these two tests are carried out.

5. The disadvantage for each of these tests is that they both increase the chance of miscarriage slightly - by up to 1% for amniocentesis and up to 2% for CVS.

Why is it important to have a low percentage of false positives in the initial screening programme?

6. The risk of Down's syndrome increases sharply with maternal age, as shown in Figure 1.

Maternal Age at Delivery	Chance of Affected Child at Term
32	1/680
34	1/475
36	1/305
38	1/190
40	1/110
42	1/65
44	1/37
46	1/21
48	1/11

Figure 1: Risk of Down's syndrome baby with increasing maternal age.

Suggest why amniocentesis and chorionic villus sampling are not routinely offered to younger women.

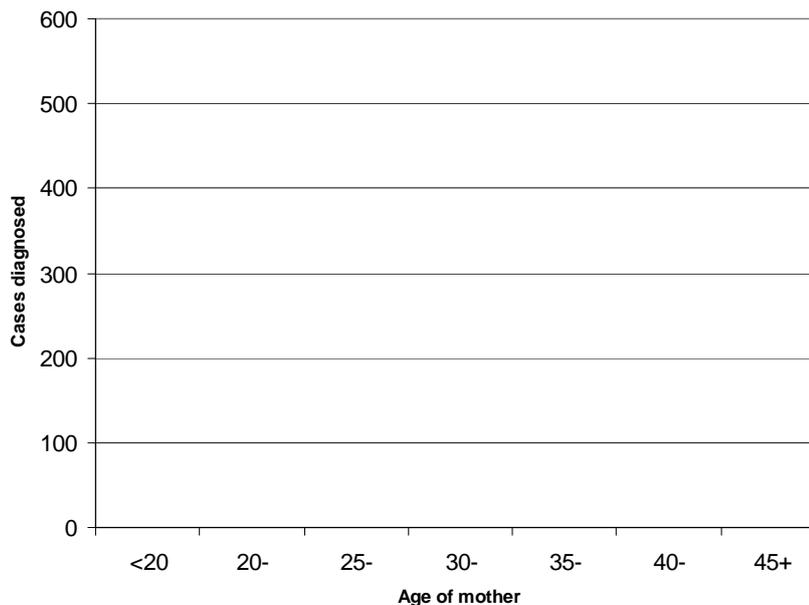
7. Some women refuse either the screening tests or the offer of further testing. Suggest two reasons for this.

8. Figure 2 gives data for the age of mothers when they were given the diagnosis of Down's syndrome using either amniocentesis or chorionic villus sampling.

Figure 2. Down's syndrome cases diagnosed in 2005 according to maternal age at diagnosis.

Maternal age (years)	No.	%
<20	28	2
20-	78	4
25-	126	7
30-	292	16
35-	568	31
40-	421	23
45+	37	2
missing	279	15
Total	1829	100

Show this data as a bar chart using the axes below.



9. Describe the trends in the data for 2005 between maternal age and diagnosis of Down's Syndrome and suggest reasons for them.
10. Currently initial screening for Down's syndrome is not compulsory for pregnant women, although it is recommended. If the NHS decided to make such screening compulsory there would be a number of ethical and financial implications that they would have to take into consideration.

Write a letter to the Health secretary explaining in detail your view on compulsory screening for Down's Syndrome.