

Genetics: screening, choice and rights

A review of current literature, websites and relevant ongoing research

Foundation for People
with Learning Disabilities

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FOREWORD

Genetic screening and testing is becoming an increasingly important issue for anyone concerned with learning disability issues. The Foundation for People with Learning Disabilities commissioned this report in order to review the research that has already been conducted on:

- the information available to pregnant women (and their partners) contemplating screening
- the availability of screening itself
- the ethical assumptions and arguments which underpin much of the basis for offering and accepting (or indeed declining) screening
- whether screening is presented and/or taken up as a genuine 'free choice'
- the implications for people with learning disabilities themselves who are contemplating screening or testing.

The authors reviewed published books and articles, grey (unpublished) research, and websites; contacted specific organisations and individuals active in this field who have undertaken or are undertaking work of particular relevance; and identified some of the key issues and themes for further discussion. (See Appendix I for further detail on this.)

The Foundation did not commission this report on the basis that all screening is automatically 'bad'; but as a body concerned with:

- improving the information and support available for people to make their own choices, and
- exploring the social attitudes and the ethical and legal framework which provide the context for those choices.

It has been very clear that although there is a considerable body of writing on some topics (notably the ethical arguments) there are other areas where there is still a lot of work to be done. We see this as a step towards exploring the implications of developments in genetics for the lives of people with learning disabilities.

Note: the text contains a number of technical terms used in genetics. Please refer to the glossary on page 17 for an explanation.

ACKNOWLEDGEMENTS

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1. INTRODUCTION: THE ISSUE

1.1 Learning disability

'Learning disability' is defined as 'a significantly reduced ability to understand new or complex information, to learn new skills (impaired intelligence), with a reduced ability to cope independently (impaired social functioning) which started before adulthood, with a lasting effect on development' (Department of Health, 2001, p14). There are many different conditions which can result in learning disability, and there is a wide range of biological, social and environmental contributory factors. There are also many people with learning disabilities who have no diagnosis of a specific condition.

The biological factors include alterations in the structure of either genes or chromosomes. Overall, it is estimated that approximately 50 per cent of all people with learning disabilities have a condition with a genetic origin (Winnepeninck et al, 2003). Of these, only half have a definite genetic diagnosis, while the remainder are presumed to have a genetic condition on the basis of clinical presentation and family history.

The two most common genetic conditions associated with the presence of learning disability are Down syndrome and fragile X (Emerson et al, 2001). Screening and testing focus predominantly at the moment on Down syndrome, and the UK National Screening Committee has recommended that all pregnant women, irrespective of age, should be offered second trimester serum screening. However, a large number of other links to single genes, multiple genes and numerical or structural chromosome anomalies have been identified (Harper, 2004). (For example, there is evidence that Klinefelter syndrome may be a common cause of learning disabilities in prepubertal boys who have previously been identified as having learning disabilities of unknown aetiology (Khalifa and Struthers, 2002), and interstitial deletions and unbalanced translocations have been detected in children with cri-du-chat syndrome (Catrinel Marinescu et al, 1999). This number will certainly increase, partly as the result of new developments in genetic science and technology (Khoury et al, 2000; Loescher and Merkle, 2005, and Peterson and Bunton, 2002) and also because a confirmed genetic contribution has now been identified for a growing number of conditions that were previously presumed to have largely environmental and/or social origins (Spinath et al, 2004).

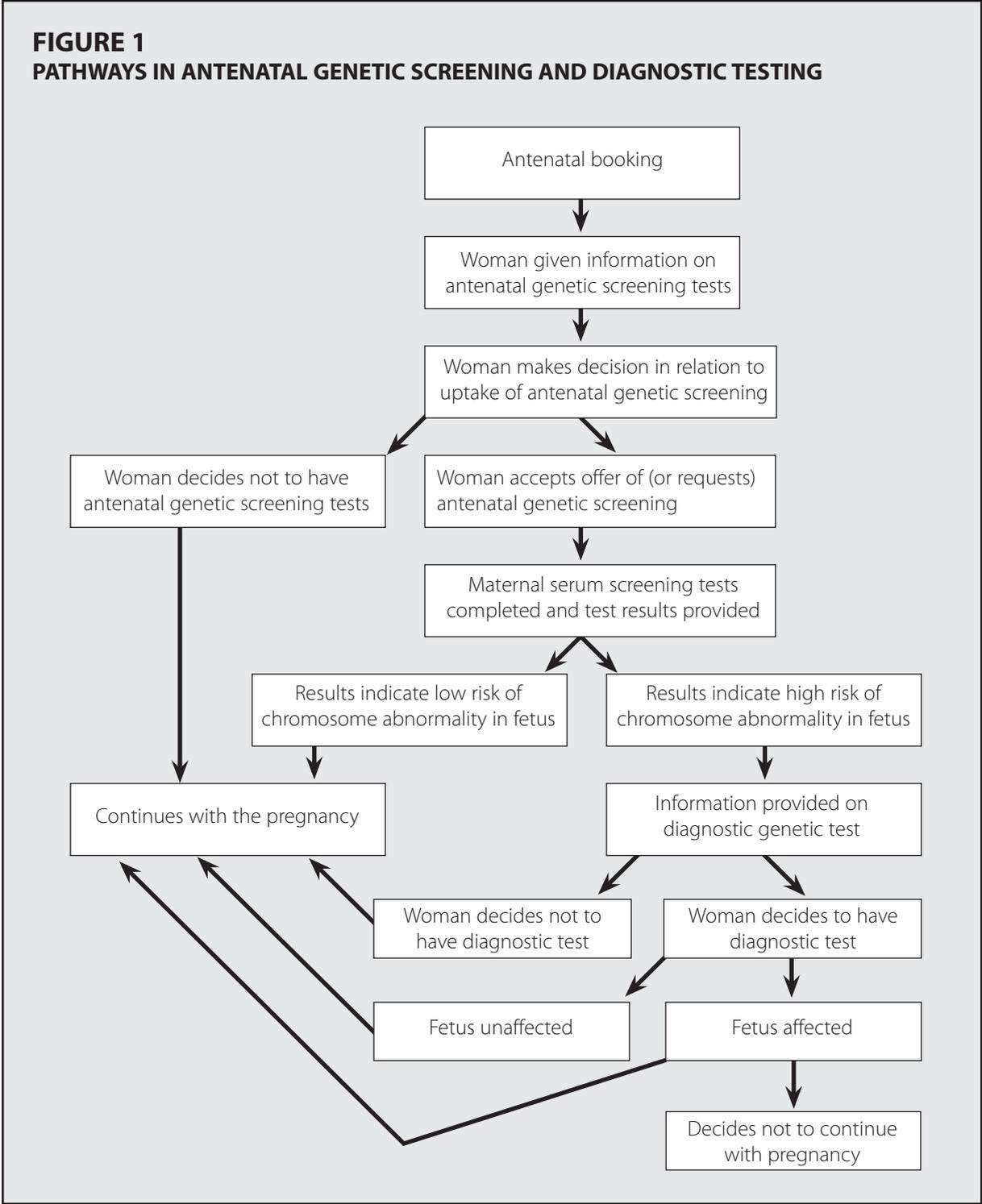
Although the current debate tends to concentrate on Down syndrome, it is clear that testing and screening are generic issues, which apply across the whole spectrum of learning disability – and that they are going to become increasingly relevant issues for pregnant women over the next decade.

1.2 Screens and tests

Although the terms are sometimes used interchangeably, there is a distinction between antenatal screening and antenatal testing. Screening involves procedures used across a general population to identify which fetuses are at higher than population risk for a genetic condition. Testing, or 'diagnostic tests', refers to the specific procedures which can give either a conclusive diagnosis or a more accurate estimation of whether this is indeed the case.

The only screening test currently on offer to all women in the UK is for Down syndrome. The most reliable screens use a combination of maternal age, biochemical test results and possibly ultrasound scan findings to devise a figure related to the chance that the fetus in that particular pregnancy may have Down syndrome (Wald et al, 1998). Laboratories may phrase screening results differently. Some use the terms 'low, moderate or high risk', while others cite the calculated numerical risk, such as a one in 200 chance that the fetus has Down syndrome. Generally those parents whose fetus is assessed as having more than a one in 250 chance are offered invasive testing (Public Health Genetics Unit, 2005). Screening for Down syndrome can also sometimes identify another type of aneuploidy (such as trisomy 13, trisomy 18 or Turner syndrome) or chromosome abnormality. Future developments in antenatal screening may include the offer of screening for fragile X (FRAX).

The pathways through the antenatal screening process, and the key decision points, are outlined in Figure 1 and additional information is provided in the flowchart produced by the National Down Syndrome Screening Programme (Appendix 2 available at http://www.nelh.nhs.uk/screening/dssp/NSC_Flowchart_2.pdf). However, the screening is not definitive, which means that some parents whose pregnancies are assessed as low risk will have a fetus with Down syndrome, while many of those who are told there is a moderate or high risk will have a normal fetus. The screening is used as guidance for offering further testing.



Women may also be offered tests because of their age, or a family history of a condition such as fragile X or Rett syndrome. Samples for testing are obtained by several means. Fetal skin cells may be harvested from a sample of amniotic fluid (obtained by amniocentesis) and are cultured to a stage in the mitotic cycle that enables the chromosome structure (karyotype) of the fetus to be examined; cells in the chorionic villi may be obtained through chorionic villus sampling (CVS) (also sometimes referred to as chorionic villus biopsy) and either viewed directly, cultured in order to examine the chromosomes or have DNA extracted; or fetal blood is taken from the umbilical cord or one of the fetal blood vessels. All these tests are performed using ultrasound guidance.

There is also a technique called Preimplantation Genetic Diagnosis (PIGD) which is used as a method of ensuring that future children of a couple do not carry a specific genetic condition for which the fetus would be at risk. This involves in vitro fertilisation, testing the blastocysts – the fertilised ova – at the 16 cell stage to see if they have the genetic mutation, and only introducing ‘normal’ blastocysts to the maternal uterus (Aittomaki et al, 2005). In England and Wales, the use of this technique is highly regulated by the Human Fertilisation and Embryology Authority (Human Fertilisation and Embryology Authority, 2005). Some people have questioned the ethical difference between PIGD and termination, although in practice it appears to have very different emotional implications for women.

1.3 The concerns

Many people, including parents of children with a learning disability, appreciate having a diagnosis of a genetic condition (Skirton, 2001; Skirton, 2006; Barr and Millar, 2003) because it enables them to plan for the future and reduces uncertainty. There is also, however, widespread concern about some of the implications for disabled people arising from developments in genetics (Avard, 2002; Holland and Clare, 2003; Soobey, 2004). These cover ethical and legal issues (Holland and Clare, 2003; Raymond, 2003; Louhiala, 2004), including the timing of tests, the process of obtaining informed and valid consent, the purpose of genetic screening and diagnostic testing, and the potential use of personal genetic information arising from test results (Human Genetics Commission, 2002).

Inevitably, there are concerns that screening and testing are being used as a way to reduce the number of disabled people – especially given the historical association with eugenics (Barr, 2002; Howarth et al, 2001; Soobey, 2004). These concerns appear to have some basis in fact, with references in the literature to the use of antenatal screening as ‘cost-effective’ (Harris et al, 2004) and as a method of reducing the number of children born with birth defects (Penchaszadeh, 2002).

There is also concern that people contemplating screening or testing are not given adequate information about what is involved, or about their options if a genetic condition linked to learning disability is diagnosed (Bryant et al, 2001b; Henley, 1996, Murray et al, 2001). Is testing a genuine, informed choice?

This report looks first at the ethical assumptions, and then at how screening is working in practice, since that practice is often influenced by the arguments.

2 ETHICS

The need to give attention to the ethical aspects of antenatal screening is highlighted in the National Down Syndrome Programme For England Handbook for Staff (UK National Screening Committee, 2004a) which states that 'the ethical implications of all screening programmes are important. These implications are particularly important in antenatal screening and the need to respect values and beliefs of different groups and individuals is of highest importance' (p.37). Most of the existing work tends to be polarised into strongly 'pro' and 'anti' screening positions. It is also not always informed (on either side) by a knowledge of disability issues or a realistic idea of the lives that people with learning disabilities lead.

At the same time, there are strong assumptions underpinning screening and information about or discussion of screening. Within the literature reviewed for this report, four key ethical arguments have been put forward to support the introduction of antenatal screening. They are summarised here – for a further detailed exploration and critique of the ethical arguments relating to the introduction of antenatal genetic testing, see Louhiala, (2004) and *Journal of Intellectual Disability Research* 47(7) which was a Special Issue on Ethics in Intellectual Disabilities and focused on genetics including antenatal screening.

2.1 'Fetal wastage'

Building from the fact that many pregnancies are lost through spontaneous abortion, and that genetic disorders are over represented in such abortions, it is argued that antenatal screening, testing and selective termination are an enhancement of a natural process. Louhiala (2004) rejects this argument: it presumes that all that happens in nature is good; it ignores the fact that plenty of fetuses with a condition leading to learning disability do survive to term; and it makes assumptions about people with a particular genotype or phenotype. It also disregards other 'natural' processes we do not try to enhance.

2.2 Parental autonomy and/or the impact on the whole family

This line of argument is based on the belief that parents, (often presented more specifically as mothers), should have the right to make autonomous decisions about their own reproduction, and that antenatal screening and diagnostic testing give them the information they are entitled to. When parents become aware that the fetus has a disability they should take this into consideration and make a decision about whether or not to continue with the pregnancy (Edwards, 2003; Louhiala, 2004). Proponents of this argument also point out that widespread screening means that younger women who were not previously offered testing for Down syndrome now receive it: conversely, older women do not need to undertake invasive tests (which can lead to spontaneous abortion (miscarriage) solely on the basis of their age, but can base their decision on the results of screening.

They also make the point that screening does not necessarily imply any particular course of action after a diagnosis. In fact, Raymond (2003) uses the example of muscular dystrophy to argue that for some parents this could reduce the frequency of termination of pregnancy, in so far as it should be possible to identify an affected fetus, rather than making the decision to terminate pregnancies of male fetuses.

A variation of this argument is that parents should have the right to take more decisions which take into consideration the effect the birth of a child with a disability may have on the lives of them as parents and other family members (Carmichael, 2003), although there is a counter argument that a termination also can have long-term consequences on the whole family (Statham, 2003). Another perspective put forward by Chadwick (2001) is that the decision to decline information and any tests should also be viewed as the exercise of autonomy and right not to know.

The view that women should have the right to make decisions over reproduction is probably the pro-screening argument that has most widespread support. However, there are still a lot of concerns about the issue of 'valid consent' (Carmichael, 2003; Edwards, 2003; Louhiala 2004 Department of Health, 2003) and about the need to balance the issue of women's autonomy against the autonomy of people with learning disabilities (Rogers and Howarth, 2001), given the fact that a diagnosis of a genetic condition is often followed by an offer of termination. Some people also point out that parental autonomy is already restricted in areas such as gender selection, and procedures aimed at genetic enhancement (Edwards, 2003; Louhiala, 2004).

The argument for parental autonomy also depends on parents having appropriate and balanced information, and the chance to make their own decisions without any pressure or compulsion. In practice, this is not always the case, as this report will explore in some detail below.

2.3 Quality of life

Another line of argument supports antenatal screening and testing on the basis that people with learning disabilities suffer as a consequence of their condition and do not have an acceptable quality of human life (Louhiala, 2004). Terminating a pregnancy therefore relieves suffering.

This assumes that all people with learning disabilities suffer, although some proponents do acknowledge that many disabled people have an excellent quality of life (Carmichael, 2003). It is also based on presumptions about what actually constitutes an acceptable quality of life, although these are often left quite vague (Edwards, 2003).

There is also the fact that, if people with learning disabilities do experience a reduced quality of life, this is not entirely (or, some people would argue, at all) the result of their genetic condition; it is the result of social barriers and the stigma currently associated with learning disabilities. Antenatal screening does not, in this context, address a key cause of the reduced quality of life among people with learning disabilities. In fact, widespread use of antenatal screening and termination of affected pregnancies have the potential to increase the stigma on those who are born with a disability.

2.4 The interest of the state

Finally, there is a belief that antenatal screening programmes are a collective issue – they are in the interest of the state. This can also be posed as an economic argument, as in the research reviews that have calculated the cost of antenatal screening programmes and used the number of births of children with Down syndrome that would need to be prevented in order for the screening programme to be cost effective (Harris et al, 2004).

Other reviews have rejected such calculations, on the basis that the purpose of antenatal screening is to promote choice rather than reduce the number of children born with disabilities (Wald et al, 1998). However, many people working in services for people with learning disabilities are particularly concerned that genetic tests and other genetic based technologies are being developed in order to reduce the number of children born with learning and other disabilities (Howarth et al, 2001; Soobey, 2004).

The counter-arguments point out that any screening programme aimed at eradicating the number of people with learning disabilities would have to become quite explicitly dysgenic, involving compulsory testing and subsequent termination, and that this would probably have limited support. In any case, given the number of non genetic causes of learning disabilities and the limitations of antenatal genetic testing to screen for all possibilities in the absence of more detailed clinical information, it would still only have a limited effect (Louhiala, 2004).

The economic calculations also assume that people with learning disabilities are economically inactive and are only a cost to society. This is increasingly not the case, since people with learning and other disabilities are entering the workforce. The estimates of the cost of caring for a person with Down syndrome also ignore the positive impact that people with learning disabilities may have on their parents, siblings and other members of society (Maxwell and Barr, 2004) as well as their contributions in paid or voluntary work.

On a more sombre note, Holland and Clare (2003) caution that, given the history of genetics and services for people with learning disabilities and previous eugenic focused approaches that had the illusion of 'scientific respectability' and were designed to 'benefit' society, we should be 'extremely cautious about compromising individual rights for the benefit of society'.

3 SCREENING AND TESTING IN PRACTICE

3.1 The recommendations

The principles of antenatal screening (Council of Europe, 1990) include:

- no screening without available pre and post test counselling
- tests should only be offered to detect serious risks to the child's health
- counselling should be non directive
- both partners should be involved if possible
- informed consent is required for screening or testing
- incapacitated persons should not be disadvantaged in terms of opportunities for screening and testing
- sufficient information should be provided for informed decisions
- pre-conceptual counselling should be offered where risks are known in advance.

The council recommends that all necessary measures are taken to ensure that, where screening or testing is offered, this does not adversely affect attitudes and behaviour towards persons who are disabled; and it also states that its recommendations are underpinned by a respect for human life and commitment to personal freedom.

The UK National Screening Committee (2003) is very clear about the information that should be provided to women considering antenatal screening. It should cover:

- the rationale for offering screening
- the fact that the screening tests are optional
- the type(s) of screening tests offered
- false positive and false negative rates
- the diagnostic tests available if results indicate the fetus is at 'high risk'
- the limitations of the screening tests
- the way in which the results will be conveyed to parents
- the options available if the fetus is diagnosed with a genetic condition, including discussion about terminating the pregnancy.

However, these guidelines do not cover such key issues as how parents would feel about having a child with a disability, or their attitudes towards termination.

The practice does not always live up to these recommendations – either in terms of information about screening, or the way that screening and its consequences are handled more generally.

3.2 Access to screening and testing across the UK

Screening policy and access to screening vary across the UK and the law relating to termination varies considerably between the different countries. All pregnant women in England, Scotland and Wales are offered antenatal genetic screening for Down syndrome and neural tube defects and have the opportunity to terminate a pregnancy if 'there is substantial risk that the child would suffer from physical or mental abnormalities as to be seriously handicapped' (Abortion Act, 1967). However, no clear definition is provided for 'seriously handicapped', although the diagnosis of Down syndrome in a fetus would be considered within this category. According to the latest available statistics, a total of 1,641 abortions were carried out in 2001 because the fetus was found to have a 'serious handicap' (National Statistics, 2001), although it is not known how many of these were on the basis of a condition associated with learning disabilities.

Northern Ireland currently has no overall antenatal screening programme. Individual Health and Social Service Trusts have local policies, which usually offer maternal serum screening tests to women over 35 years old, those with a history that suggests an increased chance of having a child with Down syndrome or women who request the test. Unlike elsewhere in the UK, prenatal diagnosis involving amniocentesis is largely co-ordinated through the Regional Clinical Genetics Services in collaboration with prenatal diagnosis clinics in two area hospitals in Northern Ireland.

3.3 Information about screening

In the United Kingdom midwives are the principal professional group providing information on antenatal screening. Women are usually given information at a routine appointment – usually (and preferably) in the early stages of the pregnancy. Midwives believe that they are the appropriate health professionals to discuss screening with women in their care (Ekelin and Crang-Svalenius, 2004).

One study indicated that midwives work very hard to ensure that clients do understand the nature of screening (Pilnick et al, 2004). However, there are some real concerns in this whole area. Samwill (2002) found that obstetricians had little confidence in midwives' knowledge of screening and midwives themselves indicated that they felt unable to offer reliable information to mothers. In a study of 63 midwives, only 26 per cent were able to answer basic questions correctly covering the parameters of the screening tests they were routinely offering to mothers (Ekelin and Crang-Svalenius, 2004). The need to invest further resources and develop consistently high quality education programmes for midwives and other professionals involved in antenatal screening has been highlighted at national level (UK National Screening Committee, 2004b).

If midwives appear to be recommending tests, many women take up the option without really thinking it through. Kuba (1995) describes the use of technology to try to obtain reassurance that the fetus is healthy as 'incredibly seductive'. One of the most revealing pieces of literature studied for this report was the paper by Pilnick (2004), which quoted a midwife who described nuchal translucency screening as 'one of the best tests we've got'. In this kind of context, many women never realise that they can and should make active choices about antenatal screening.

Midwives are also, in practice, being expected to support women in decisions about screening and termination which can challenge them both personally and morally (Wray, 2001). Ekelin and Crang-Svalenius (2004) discovered that midwives find this aspect of their work extremely difficult; a number in that study said they were relieved not to have to make similar decisions for themselves.

3.4 Assumptions about learning disability

The professional literature does not contain explicitly negative portrayals of people with learning disabilities, but there is an underlying assumption that Down syndrome or other conditions would by definition be a 'bad thing'. For example, Hey and Hurst (2003) title their article on uptake of antenatal screening 'Antenatal screening: why do women refuse?' and cite a midwife who says that giving information in the 'right way' means few women will refuse screening tests. Other research studies aim to identify methods for increasing the uptake of screening (Michie et al, 2004).

Alderson (2001) points to a more general ignorance of learning disability throughout the health services. This ignorance – and fear of the unknown – influences the information and implicit messages that parents receive; Fletcher (2001) argues that at times this is evident in the language such as 'bad genes', 'cruel inheritance' and 'faulty genes'.

3 Screening and Testing in Practice

This ignorance often extends to professionals offering screening and to the people undergoing screening themselves. As the number of terminations for Down syndrome increases, the opportunities reduce for individuals to meet and interact with children and adults who have Down syndrome. Blumberg (1994) holds the view that genetic counsellors have a skewed impression of individuals with learning disabilities because they most often meet those who are experiencing difficulties at the extreme end of the spectrum. Midwives' knowledge of Down syndrome and the lives of people with Down syndrome has also been questioned (Samwill, 2002; Williams et al, 2002b).

Parents are being asked to make a decision about something of which they may have no direct experience; by extension, they are not able to give actual truly informed consent to these procedures (Bromberg Bar-Yam, 2003). It may be relevant to note that research to explore the views of women who have a sibling with Down syndrome indicates that only about half (54 per cent) would definitely wish to have an antenatal diagnostic test during a pregnancy, and some were distressed and offended by the health professionals' assumptions that they would want it.

4 PEOPLE WITH LEARNING DISABILITIES

Most of the existing literature focuses on the ethical issues (Holland and Clare, 2003). There has been much less of a focus on evaluating the actual implementation of antenatal screening, and the actual or potential practical implications for parents, other family members and people with learning disabilities.

The past decade has seen an emphasis on providing accessible information to people with learning disabilities about a wide range of aspects of health care and equity of access to services across the United Kingdom and internationally (Scottish Executive, 2000; Department of Health, 2001; Department of Health, Social Services and Public Safety, 2004; Sowney and Barr, 2004). Women with learning disabilities do become pregnant and use maternity services, so they require accessible information (Department of Health, 2001). However, in reviewing the available literature it became very clear that few attempts have been made to date to explain the concepts of genetics to people with learning disabilities, or explore what people with learning disabilities understand and feel about genetics in general and antenatal screening in particular.

Ward (2001) and her colleagues have demonstrated that individuals with learning disabilities are clearly able to understand the issues around antenatal screening and testing if they are properly informed about them. However, the literature review uncovered no specific information packages on antenatal screening for people with learning disabilities or specific information for people with sensory impairments; if they do exist, they are extremely hard to find. This is clearly a major gap in the current literature, given the widely acknowledged importance of informed consent and valid decision making.

In an innovative project Howarth et al (2001) sought to provide information about genetics, including antenatal screening and termination, to a group of people with learning disabilities. This was provided in a workshop session and using specially formatted information that sought to explain the concepts involved. It was clear from the feedback of the people with learning disabilities that they understood the key points and recognised that antenatal screening could lead to termination if tests confirmed Down syndrome. Although they acknowledged some difficulties in living with learning disabilities, they also highlighted their achievements; and several, but not all, people questioned why the birth of a child with disabilities should be prevented. One person with Down syndrome became particularly distressed at the realisation that the birth of people with Down syndrome could be prevented. Alderson et al (2001) reported a similar distress among some people with Down syndrome she interviewed in relation to genetics and she notes the need for supportive counselling in providing this information – a criterion already expected when providing this information to people who do not have learning disabilities (UK National Screening Committee, 2004a).

5 CHOICE IN CONTEXT

How far do pregnant women and their partners have a genuine choice over whether to opt for screening, or on how to act on the results of a test that confirms a genetic condition associated with learning disability?

Many researchers cite the fact that antenatal screening has become 'routine', so that women may not realise they have a choice to make and a right to decline (Blumberg 1994; Press and Browner, 1997; Pilnick, 2004; Tsianakas and Liamputtong, 2002a). Anderson (1999) asserts that, because in our society science is seen as 'a good thing' and professionals are assumed to have altruistic motives, the offer of genetic testing is perceived as a directive to be followed rather than an informed parental choice. Benzie et al (2004) investigated women's knowledge of antenatal screening tests and concluded that women regarded screening as part of an unquestioned routine; on the other hand, Kaiser et al (2004), reported that pregnant women face difficult decisions at every step in the antenatal screening process.

Uptake rates for antenatal screening vary between countries. The reasons for accepting or declining are often based on women's personal values and views towards termination (van den Berg, 2005). Similar variation has recently been reported among sisters of people with Down syndrome. However, there is also evidence that those women who did accept antenatal screening are motivated by a desire to reduce anxiety about the health of the fetus, rather than because they wish to discover if the fetus has an abnormality (Sturm and Ormond, 2004). Others feel that they are doing their best for the baby by taking up the offer of screening (Tsianakas and Liamputtong, 2002b). Bryant et al (2005) found that while the majority (54 per cent) of women in their study of 78 women reported they would use diagnostic tests, a similar majority (53 per cent) would not consider termination on the grounds of Down syndrome.

When it comes to results, explanations of the screening process that focus on the genetic aspects rather than on outcomes in psychosocial terms may influence couples to think of the outcomes in terms of medical abnormality to which the solution is a termination (Alderson, 2001). In addition, anyone interpreting the screening results needs to understand the concept of risk. This is not always the case (Gates, 2004), and risk is often explained in terms of pathophysiology, rather than in terms of ensuring that women who want to continue with their pregnancies can do so (Hunt et al, 2005). In her personal story, Kuba (1995) describes her fury at being reminded repeatedly that she could opt for a termination, after she had clearly expressed her wish to continue with the pregnancy.

These reports highlight the discrepancy between professional and patient agendas, and the resulting poor communication and lack of supportive care. Furthermore, there are concerns that parents who knowingly choose to continue with a pregnancy after a congenital disorder has been diagnosed may be ostracised or made to feel guilty for that decision (Driver, 2004; Wyatt, 2000). In a study of nursing attitudes, nurses were more likely to blame parents for their child's disability in cases that could have been detected antenatally (Sharu, 1996). A survey conducted for the Down's Syndrome Association in May 2006 found that 22 per cent of those parents who had decided to go ahead with a pregnancy which they knew would mean giving birth to a baby with Down Syndrome felt their decision was not supported by health professionals.

The assertion that termination is chosen to reduce pain and suffering for the child is contradicted by the findings of Bell and Stoneman (2000). In that study, respondents were more willing to terminate a pregnancy on the basis of Down syndrome than spina bifida or haemophilia, both conditions that may seriously impair the health of the child and may require long-term medical treatment. It would be highly relevant to discover the reasoning behind these responses; it may be that parents feel that a learning disability would be more incapacitating than a physical one, or that they believe that people with Down syndrome suffer on the basis of their disability.

However, relevant studies indicate that women are on the whole satisfied with their ability to make informed choices. O'Caithlin et al (2002) studied women who had been offered antenatal screening, 73 per cent of whom believed that they had been able to make an informed choice. Similar findings were reported by Bulman et al (2004); 75 per cent of women in that study had serum screening, with 88.7 per cent agreeing that they had a choice whether to have screening and 81.7 per cent believing they were involved in the decision making process.

6 RECOMMENDATIONS

This report was commissioned to investigate the existing literature and information on genetic screening and its implications. Its findings highlight:

- the patchy information and support available to members of the public (ie pregnant women and their partners, either before or after any kind of screening or testing)
- the gap between the principles and practice
- and the lack of basic information about the lives of those born with a disability such as Down syndrome.

A lot of work has been published on the ethical arguments, and some services for people with learning disabilities have been involved in public consultation on this issue. However, there are still real concerns about the information surrounding screening, and the assumptions about learning disability which underpin this information.

We need a clearer picture of exactly what information women receive when they are contemplating screening, and the way in which this information is presented. However, it seems fairly clear that what is currently on offer does not consistently meet the guidelines.

This look at the existing research should form the basis of a more rigorous investigation into the experiences of prospective parents (with and without learning disabilities) and the staff conducting screening. Monitoring of the standards that have already been developed for the antenatal screening programme should also produce some useful data. However, these standards have their limitations, as they specify neither further information on the lives of people with learning disabilities, nor information accessible to people with learning disabilities.

Given those limitations, there is also an urgent need to establish the views of people with learning disabilities, including people with Down syndrome and their families, towards antenatal genetic screening and testing; and to establish just how much realistic, up to date information is available to professionals and to prospective parents about people with learning disabilities and their lives.

This investigation could lead to proposals for improving the information and training on offer, and the standards themselves.

7 CONCLUSION

The authors who researched this report found that doing so made them reflect on their values, beliefs and professional practice. Skirton says, 'When I began this piece of work, I brought to it my experiences as a paediatric nurse, as a midwife and as a genetic counsellor. In those roles I had seen myself as an advocate for patients and their families, and as a believer in choices for parents. I was familiar with much of the literature described in the report. However, I have been faced with questions about the reality of choice for parents. It is particularly disturbing to realise the extent to which screening has become "routinised" and the challenges faced by parents who wish to decline screening. In addition, terms I once thought reasonably neutral, such as "high risk fetus", appear to me now to carry a negative connotation that could influence parental choice and attitudes towards disabled people.'

Barr, who has a background in learning disability nursing, adds, 'I thought I had clear ideas about the rights of people with learning disabilities and beliefs about how I viewed antenatal screening. However, I have found myself challenged and at times troubled by seeking to reconcile the rights of people to choice (including people with learning disabilities who do not wish to have children with learning disabilities) and the rights of people with learning disabilities.'

Genetic screening is a complex and contentious issue and balancing the 'choice' agenda against the 'rights' one is profoundly difficult for anyone who feels that both views have validity. This report demonstrates clear needs for:

- better informed debate
- better information and processes for prospective parents
- stronger standards and training for practitioners.

As genetic research and technology progress, these needs will become even more pressing than they are today.

GLOSSARY

Affected individual: A person who has the signs and symptoms of the genetic condition.

Amniocentesis: Withdrawal of amniotic fluid from the amniotic sac, usually for the purpose of testing the fetal chromosomes.

Anencephaly: Failure of the anterior neural tube to close properly during very early intrauterine life, resulting in the absence of the cerebral hemispheres and skull bone, and a rudimentary brain stem.

Aneuploidy: An alteration in the number of chromosomes, involving only one or several chromosomes rather than the entire set of chromosomes.

Assisted reproduction: Any artificial technique used to enable a pregnancy to be achieved (e.g. in vitro fertilisation).

Carrier: A person who is generally not affected with the condition, but carries one mutated copy of a gene. Generally relates to heterozygotes in recessive or X-linked conditions.

Chorionic villus biopsy: Removal of cells from the chorionic villi (developing placental tissue).

Chromosome: The physical structures into which the DNA is packaged within the nucleus of cells. The usual number of chromosomes in humans is 46.

Clinical genetics: The branch of the health service that is chiefly involved in diagnosis of genetic conditions and genetic counselling for families.

Cordocentesis: Removal of a sample of fetal blood from the umbilical cord during pregnancy.

Cytogenetics: The study of chromosomes, in the laboratory.

DNA: Deoxyribonucleic acid. The biochemical substance that forms the genome. It carries in coded form the information that directs the growth, development and function of physical and biochemical systems. It is usually present within the cell as two strands with a double helix conformation.

Dysmorphic features: Physical features that are outside of the variability of the normal population. They may occur because of a change in the genetic code providing instructions for those features.

Eugenics: The study and practice of principles which aim to 'improve' the genetic status of a population in line with a stated belief in 'health and fitness'.

Gene therapy: Therapy that is based upon the principle of replacing or modifying a faulty gene in the relevant tissues. The aim is to reduce or obliterate the effects of the genetic condition.

Gene: The fundamental physical and functional unit of heredity consisting of a sequence of DNA.

Genetic counsellor: A person whose main professional role is to offer information and support to clients who are concerned about a condition that may have a genetic basis.

Genetic screening: This term usually refers to population screening for a genetic variation or mutation.

Genomics: the study of interactions between genetic and environmental factors that contribute to disease.

Genotype: the genetic makeup of an individual or the specific gene structure at one locus.

Induced abortion: Termination of pregnancy.

Karyotype: A description of the chromosome structure of an individual (assessed during metaphase), including the number of chromosomes and any variation from the normal pattern.

Maternal serum screening: A method of detecting a relative risk of Down syndrome, some other chromosomal abnormalities and neural tube defects in a pregnancy, using biochemical testing of the mother's blood.

Mendelian disorder or Mendelian condition: A genetic disorder caused by a single gene mutation, following a dominant, recessive or X-linked pattern of inheritance.

Mutation: A gene sequence variation that is found in less than one per cent of the total population. The mutation may cause a change in the protein product of the gene, and therefore cause health problems for the person concerned.

Neural tube defect: An abnormality of the spinal column or cranium (spina bifida or anencephaly).

Non-directiveness: A model of counselling used in genetic counselling, which emphasises the right of clients to make decisions without coercion from others.

Pedigree: Family tree.

Phenotype: the clinical manifestation (signs and symptoms) of the condition

Population screening: Using a test to assess the risk or presence of a disorder in an entire section of the population e.g. neonatal screening for hypothyroidism.

Proband: The affected person in the family or the person who is seeking genetic advice.

Recurrence risk: The chance that a genetic condition will occur again in offspring or siblings of an affected person.

Spina bifida: An interruption to the spinal column, with possible herniation of the spinal cord and meninges (myelomeningocele). One form of neural tube defect (another being anencephaly).

Spontaneous abortion: Loss of a pregnancy without interference, miscarriage.

Syndrome: A number of physical features or abnormalities that fit a recognised pattern.

Trisomy: Having three copies of a particular chromosome.

Ultrasound scanning: Investigation of physical structures using ultrasound device (sound waves).

REFERENCES

Abortion Act (1967). <http://members.aol.com/abtrbng/aa67.htm> Accessed 28 June 2005.

Aittomaki K, Bergh C, Hazekamp J, Nygren KG, Selbing A, Soderstrom-Anttila V, Wennerholm UB. (2005) Genetics and assisted reproduction technology.

Acta Obstetrics and Gynecology Scandinavia, 84(5):463-73.

Aksoy S. (2001) Antenatal screening and its possible meaning from an unborn baby's perspective. *BMC Medical Ethics*, 2:3. doi: 10.1186/1472-6939-2-3.

Alderson P, Aro AR, Dragonas T, Ettorre E, Hemminki E, Jalinoja P, Santalahti P, Tymstra T. (2001) Prenatal screening and genetics. *European Journal of Public Health*, 11(2):231-3.

Alfirevic Z, Neilson JP. (2004) Antenatal screening for Down's syndrome. *British Medical Journal*, 329(7470):811-2

Anderson G. (1999) Nondirectiveness in prenatal genetics: patients read between the lines. *Nursing Ethics*, 6(2):126-36.

Avard D. (2002) Human Genetics Research and Practice: Implications for People with Disabilities. Report of the United Nations expert group meeting on disability-sensitive policy and programme monitoring and evaluation. UNHQ, New York, 3-5 December, 2001.

Bane A, Brown L, Carter J, Cote C, Crider K, de la forest S, Livingston M, Montero D. (2003) Life and Death Decisions: America's Changing Attitudes Towards Genetic Engineering, Genetic Testing and Abortion, 1972-98 *International Social Work*, 46(2):209-219

Barr, O. (2002) Developments in genetic understanding: time to engage with the process. *Journal of Learning Disabilities*, 6(1): 5-12.

Barr O, Millar R. (2003) Parents of children with intellectual disabilities: their expectations and experience of genetic counselling. *Journal of Applied Research in Intellectual Disabilities*, 16(3):189-204.

Bell M, Stoneman Z. (2000) Reactions to Prenatal testing: Reflection of Religiosity and Attitudes Toward Abortion and People with Disabilities. *American Journal on Mental Retardation*, 105(1):1-13.

Benn PA, Egan JF, Fang M, Smith-Bindman R. (2004) Changes in the utilization of prenatal diagnosis. *Obstetrics and Gynecology*, 103(6):1255-60.

Benzie RJ, Kennedy N, Martin R, Mein B, Miceli F, Thavaravy R, Webb A. (2004) How much do women know about first trimester ultrasound and serum screening? *ASUM Ultrasound Bulletin*, 7(3):13-14.

Blumberg L. (1994) The politics of Prenatal Testing and Selective Abortion. *Sexuality and Disability*, 12(2):135-53.

Bromberg Bar-Yam N. (2003) Prenatal Testing: Personal Choices and Social Context. *International Journal of Childbirth Education*, 18(3):18-20.

- Bryant, LD., Murray, J., Green, JM., Hewison, J., Sehmi, I., Ellis, A.,** (2001a) Descriptive information about Down syndrome: a content analysis of serum leaflets. *Prenatal Diagnosis*, 21:1057-1063.
- Bryant LD, Green JM, Hewison J.** (2001b) Prenatal screening for Down's syndrome: some psychosocial implications of a 'screening for all' policy. *Public Health*, 115(5):356-8.
- Bryant L, Hewison JD, Green JM.** (2005) Attitudes towards prenatal diagnosis and termination in women who have a sibling with Down's syndrome. *Journal of Reproductive and Infant Psychology*, 23(2):181-198.
- Bulman J, Nan A, Markland MH, Al-Safi W, Leeson S.** (2004) A questionnaire study of women's views regarding the effectiveness of serum Down screening in north west Wales. *Journal of Obstetrics and Gynaecology*, 24(1):33-9
- Burn J, Fairgrieve S, Franks P, White I, Magnay D.** (1996) Audit of maternal serum screening: strategies to augment counselling in response to women's views. *European Journal of Human Genetics*, 4(2):108-12.
- Canick J.** (2003) Safety first: choices in antenatal screening for Down's syndrome. *Journal of Medical Screening*, 10(2):55.
- Carmichael, B., Pembery, M., Turner, G., Barnicoat, A.** (1999) Diagnosis of Fragile-X syndrome: the experiences of parents. *Journal of Intellectual Disability Research*, 43(1):47-53.
- Carmichael B.** (2003) The Human Genome Project- threat or promise? *Journal of Intellectual Disability Research*, 47(7):505-508.
- Chadwick R.** (2001) Whose choice? Whose responsibility? Ethical issues in prenatal diagnosis and learning disability. In Ward L. (Ed.) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities* (pp.82-91). Kidderminster: British Institute of Learning Disabilities.
- Chapple, A., May, C.** (1996) Genetic knowledge and family relationships: two case studies. *Health and Social Care in the Community*, 4(3):166 - 171.
- Collins FS, McKusick VA.** (2001) Implications of the Human Genome Project for medical science. *Journal of the American Medical Association AMA.*, 285(5):540-4.
- Collins, V., Halliday, J., Kahler, S., Williamson, R.** (2001) Parents' experiences with genetic counseling after the birth of a baby with a genetic disorder: an exploratory study. *Journal of Genetic Counseling*, 10(1): 53-72.
- Council of Europe** (1990) Council of Europe, Committee of Ministers, Recommendation No. R (90) 13 on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling (June 21, 1990), <http://www1.umm.edu/humanrts/instree/coerecr90-13.html> Accessed 10 June 2005.
- de Graaf IM, Tijmstra T, Bleker OP, van Lith JM.** (2002) Womens' preference in Down syndrome screening. *Prenatal Diagnosis*, 22(7):624-9.
- Department of Health** (2001) *Valuing People. A new strategy for learning disability for 21st century*, Cm 5086. London: Department of Health.

Department of Health (2003) *Our Inheritance, Our Future: Realising the potential of genetics in the NHS Cm 5791-II*. London: The Stationary Office.

Department of Health, Social Services and Public Safety (2004) *Equal Lives: Draft report of Learning Disability Committee*. Belfast: Department of Health, Social Services and Public Safety.

Dimivicus J. (1997) Information giving and decision making in antenatal screening. www.prochoiceforum.org.uk/and2asp Accessed 10 June 2005.

Driver D. (2004) Genetic Screening: Is it ethical? *BioNews*. <http://www.bionews.org.uk/commentary.lasso?storyid=2300> Accessed 10 June 2005.

Edwards SD. (2003) Prenatal screening for intellectual disability. *Journal of Intellectual Disability Research*, 47(7):526-32.

Ekelin M, Crang-Svalenius E. (2004) Midwives' attitudes to and knowledge about a newly introduced fetal screening method. *Scandinavian Journal of Caring Sciences*, 18(3):287-93

Emerson E, Hatton C, Felce D, Murphy G. (2001) *Learning Disabilities: the fundamental facts*. London: The Foundation for People with Learning Disabilities.

Fairgrieve S, Franks P, White I, Magnay D. (1996) Audit of maternal serum screening: strategies to augment counselling in response to women's views. *European Journal of Human Genetics*, 4(2):108-12.

Fletcher A. (2001) 'Three generations of imbeciles are enough': eugenics, the new genetics and people with learning difficulties. In Ward L. (Ed.) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities* (pp.72-81). Kidderminster: British Institute of Learning Disabilities.

Gates EA. (2004) Communicating Risk in Prenatal Genetic Testing. *Journal of Midwifery and Women's Health*, 49(3):220-7.

Green JM, Hewison J, Bekker HL, Bryant LD, Cuckle HS. (2004) Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review. *Health Technology Assessment* 8(33):iii, ix-x, 1-109.

Harris RA, Washington AE, Nease RF Jr, Kuppermann M. (2004) Cost utility of prenatal diagnosis and the risk-based threshold. *Lancet*, 363(9405):276-82.

Harper PS. (2004) *Practical Genetic Counselling 6th Edition*. Oxford, Oxford University Press. (replaces an earlier edition)

Henley A. (1996) *Pregnancy and Antenatal Care*. Chapter 19 in *Culture, religion and childbearing in a multiracial society*. Judith Schott and Alix Henley. Oxford: Butterworth-Heinemann.

Hey M, Hurst K. (2003) Antenatal screening: why do women refuse? *Midwives*, 6(5):216-20.

Holland A, Clare ICH. (2003) The Human Genome Project: considerations for people with intellectual disabilities. *Journal of Intellectual Disability Research*, 47(7):514-525.

Howarth, J., Rodgers, J., Collins, A., Cook, B., Hamblett, G., Harris, C., Long, J., May, Z., Webster, B. (2001) Difference and choice: a workshop for people with learning disabilities. In Ward, L. (Ed.) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities* (pp.27-42). Kidderminster: British Institute of Learning Disabilities.

Human Fertilisation and Embryology Authority (2005)
www.hfea.gov.uk/HFEAGuidance Accessed 23 June 2005.

Human Genetics Commission (2005) *Profiling the newborn: a prospective gene technology?* London: Department of Health.

Human Genetics Commission (2002) *Inside information: balancing interests in the use of personal genetic data.* London: Department of Health.

Hunt LM, de Voogd KB, Castaneda H. (2005) The routine and the traumatic in prenatal genetic diagnosis: does clinical information inform patient decision-making? *Patient Education and Counselling*, 56(3):302-12.

Kaiser AS, Ferris LE, Katz R, Pastuszak A, Llewellyn-Thomas H, Johnson J, Shaw BF. (2003) Psychological responses to prenatal NTS counselling and the uptake of invasive testing in women of advanced maternal age. *Patient Education and Counseling*, 54:45-53.

Khoury M., Burke W. & Thompson, J. (Eds.) (2000) *Genetics and public health in the 21st century: using genetic information to improve health and prevent disease.* New York: Oxford University Press.

Kornman L, Chambers H, Nisbet D, Liebelt J. (2002) Pre-conception and antenatal screening for the fragile site on the X-chromosome. *Cochrane Database Systematic Review*, (1):CD001806.

Kuba LM. (1995) The Prenatal Testing Roller Coaster: One Mother's Story. *The Journal of Perinatal Education*, 4(4):19-22.

Lejeune J, Gautier M. and Turpin, R. (1959). Mongolism; une maladie chromosomique (trisomy) [Mongolism; a chromosomal disease (trisomy)]. *Bulletin of the Academy of National Medicine*, 143, 256-265.

Leung KY, Lee CP, Chan HY, Tang MH, Lam YH, Lee A. (2004) Randomised trial comparing an interactive multimedia decision aid with a leaflet and a video to give information about prenatal screening for Down syndrome. *Prenatal Diagnosis*, 24(8):613-8.

Louhiala P. (2004) *Preventing Intellectual Disability. Ethical and Clinical issues.* Cambridge: Cambridge University Press.

McConkie-Rosell A, Spiridigliozzi, GA, Sullivan JA, Dawson DV, Lachiewicz AM. (2000) Carrier testing in fragile-X syndrome: effect on self concept. *American Journal of Medical Genetics*, 92:336-342.

McLaughlin J. (2003) Risky professional boundaries. *Articulations of the personal self by antenatal screening professionals.* *Journal of Health Organisation and Management*, 17(4):264-79.

Maxwell V, Barr O. (2003) With the benefit of hindsight: a mother's reflection on raising a child with Down Syndrome. *Journal of Learning Disabilities*, 7(1): 51-64.

- Michie S, Dormandy E, Marteau TM.** (2003) Informed choice: understanding knowledge in the context of screening uptake. *Patient Education and Counselling*, 50(3):247-53.
- Michie S, Dormandy E, Marteau TM.** (2004) Increasing screening uptake amongst those intending to be screened: the use of action plans. *Patient Education and Counselling*, 55(2):218-22.
- Mulvey S, Pham T, Tyzack K, Wallace EM.** (2002) Women's preferences for reporting of Down syndrome screening results. *Australia and New Zealand Journal of Obstetrics and Gynaecology*, 42(5):504-7
- Munim S, Khawaja NA, Qureshi R.** (2004) Knowledge and awareness of pregnant women about ultrasound scanning and prenatal diagnosis. *Journal of Pakistani Medical Association*, 54(11):553-5.
- Murray, J., Cuckle, H., Sehmi, I., Wilson, C., Ellis, A.** (2001) Quality of written information in Down syndrome screening. *Prenatal Diagnosis*, 21:138-142.
- National Statistics** (2001) Series AB No 28 Abortion Statistics. www.nationalstatistics.gov.uk/downloads/theme_health/AB28_2001/AB28_2001.pdf London, The Stationery Office. Accessed 16.6.05
Burn J, www.un.org/esa/socdev/enable/disid2001e.htm Accessed 7 April, 2005.
- O’Cathain A, Thomas K, Walters SJ, Nicholl J, Kirkham M.** (2002) Women's perceptions of informed choice in maternity care. *Midwifery*, 18: 136-44.
- Penchaszadeh VB.** (2002) Preventing congenital anomalies in developing countries. *Community Genetics*, 5(1):61-9.
- Petersen A, Bunton R.** (Eds.) (2002) *The new genetics and the public's health*. London: Routledge.
- Pilnick A.** (2004) 'It's just one of the best tests that we've got at the moment': the presentation of nuchal translucency screening for fetal abnormality in pregnancy. *Discourse and Society*, 15(4):451-65.
- Pilnick AM, Fraser DM, James DK.** (2004) Presenting and discussing nuchal translucency screening for fetal abnormality in the UK. *Midwifery*, 20(1):82-93.
- Press N, Browner CH.** (1997) Why women say yes to prenatal diagnosis. *Social Science and Medicine*, 45(7):979-89.
- Public Health Genetics Unit** (2005)
www.phgu.org.uk/info_database/diseases/downs_syndrome/downs.html Accessed 23 June 2005.
- Raymond FL.** (2003) Genetic services for people with intellectual disability and their families. *Journal of Intellectual Disability Research*, 47(7): 509-514.
- Reinders H.** (2000) *The future of the disabled in Liberal society: an ethical analysis*. Indiana: University of Notre Dame Press.
- Richards M.** (1996) Lay and professional knowledge of genetics and inheritance. *Public Understanding of Science*, 5: 217-230.

Rioux M. (2001) The many interest in genetic knowledge: an international perspective on prenatal screening and the use of genetic information in relation to people with learning difficulties. In Ward L. (Ed.) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities* (pp.91-107). Kidderminster: British Institute of Learning Disabilities.

Rodgers J, Howarth J. (2001) Difference and choice: helping people with learning difficulties to consider ethical issues around genetics. In Ward L. (Ed.) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities* (pp.16-26). Kidderminster: British Institute of Learning Disabilities.

Samwill L. (2002) Midwives' knowledge of Down's syndrome screening. *Br J Midwifery*, 10(4):247-50

Scottish Executive (2000) *The same as you? A review of the services for people with learning disabilities.* Edinburgh: Scottish Executive.

Sharu D. (1996) Attribution of blame for a child's disability. *Professional Nurse*, 11(12):790-2.

Shaw C, Abrams K, Marteau T. (1999) Psychological impact of predicting individual's risk of illness: a systematic review. *Social Science and Medicine*, 49:1571-1598.

Skirton H. (2001) The Client's Perspective of Genetic Counseling – A Grounded Theory Study. *Journal of Genetic Counseling*, 10(4):311-29.

Skirton H, Patch C. (2002) *Genetics for healthcare professionals.* Oxford: BIOS.

Skirton H. (2006) Parental experience of a pediatric genetic referral. *MCN American Journal of Maternal and Child Nursing*. 31(3): 178-184

Song FJ, Barton P, Sleightholme V, Yao GL, Fry-Smith A. (2003) Screening for fragile X syndrome: a literature review and modelling study. *Health Technology Assessment*, Vol 7: No 16.

Soobey RD. (2004) Valuing People with learning disabilities in the context of the Human Fertilisation and Embryology Act 1990: social policy and legislative incompatibility. *Journal of Learning Disabilities*, 8(2):107-114.

Spencer K, Aitken D. (2004) Factors affecting women's preference for type of prenatal screening test for chromosomal anomalies. *Ultrasound Obstetrics and Gynecology*, 24(7):735-9

Spinath F, Harlaar N, Ronald A, Plomin R. (2004) Substantial genetic influence in mild mental impairment in early childhood. *American Journal of Mental Retardation*, 109 (1): 34-43.

Stainton T. (2003) Identity, difference and the ethical politics of prenatal testing. *Journal of Intellectual Disability Research*, 47(7):533-9.

Statham H. (2002) Prenatal diagnosis of fetal abnormality: the decision to terminate the pregnancy and the psychological consequences. *Fetal and Maternal Medicine Review*, 13(4):213-47.

Sturm EL, Ormond KE. (2004) Adjunct Prenatal Testing: Patient Decisions Regarding Ethnic Carrier Screening and Fluorescence In Situ Hybridization. *Journal of Genetic Counseling*, 13(1):45-63.

Tsianakas V, Liamputtong P. (2002a) What women from an Islamic background in Australia say about care in pregnancy and prenatal testing. *Midwifery*, 18:25-34.

Tsianakas V, Liamputtong P. (2002b) Prenatal testing: the perceptions and experience of Muslim women in Australia. *Journal of Reproductive and Infant Psychology*, 20(1):7-24.

Tyzack K, Wallace EM. (2003) Down syndrome screening: what do health professionals know? *Australia and New Zealand Journal of Obstetrics and Gynaecology*, 43(3):217-21.

UK National Screening Committee (2003) Antenatal screening – working standards incorporating those for the National Down Syndrome screening programme for England. www.nelh.nhs.uk/screeningdssp/programme.html Accessed 23 June 2005

UK National Screening Committee (2004a) Antenatal screening – working standards incorporating those for the national Down Syndrome screening programme for England. Kettering. UK National Screening Committee.

UK National Screening Committee (2004b) National Training Needs Analysis for antenatal screening across England. Kettering. UK National Screening Committee.

UK National Screening Committee (2005) www.nelh.nhs.uk/screening/dssp/NSC_Flowchart_2.pdf. Accessed 23 June 2005.

UK National Screening Committee (2005) DoSySp: The newsletter of the National Down syndrome screening programme. Issue 2: April 2005. Kettering. UK National Screening Committee.

van den Berg M, Timmermans DRM, Kleinveld JH, Garcia E, van Vugt JMG, van der Wal G. (2005) Accepting or declining the offer of prenatal screening for congenital defects: test uptake and women's reasons. *Prenatal Diagnosis*, 25:84-90.

Vause S, Sands J, Johnston TA, Russell S, Rimmer S. (2002) Could some fetocides be avoided by more prompt referral after diagnosis of fetal abnormality? *Journal of Obstetrics and Gynaecology*, 22(3):243-5.

Wald NJ, Kennard A, Hackshaw A, McGuire A. (1998) Antenatal screening for Down's syndrome. *Health Technology Assessment*, Vol 2 No 1. <http://www.nchta.org/execsumm/summ201.htm> Accessed 10 June 2005.

Wald NJ, Huttly WJ, Hackshaw AK (2003). Antenatal screening for Down's syndrome with the quadruple test. *Lancet*, 361:835-836.

Ward L. (Ed.) (2001) *Considered Choices? The new genetics, prenatal testing and people with learning disabilities*. Kidderminster: British Institute of Learning Disabilities.

Williams C, Sandall J, Lewando-Hundt G, Heyman B, Spencer K, Grellier R. *Women as moral pioneers?* (2005) Experiences of first trimester antenatal screening. *Social Science and Medicine* May 14; [Epub ahead of print]

Williams C, Alderson P, Farsides B. (2002a) Is nondirectiveness possible within the context of antenatal screening and testing? *Social Science and Medicine*, 54:339-347.

Williams, C., Alderson, P., Farsides, B. (2002b) What constitutes 'balanced information in the practitioners' portrayal of Down syndrome. *Midwifery*, 18:230-237.

Winnepenninck B, Rooms L, Kooy R. (2003) Mental retardation: a review of genetic causes. *British Journal of Developmental Disabilities*, 49(1):29-44.

World Health Organisation (2005) Antenatal Screening and Birth Defects Surveillance. <http://www.afro.who.int/drh/antenatal.html> Accessed 10 June 2005.

Wray J. (2002) Antenatal screening for Down's syndrome and the midwife: midwives views count? www.fhsc.salford.ac.uk/hcprdu/events/midwives.htm Accessed 10 June 2005.

Wyatt J. (2000) The New Ethics of Abortion. *Triple Helix*, Summer 2000. P11-13.

APPENDIX I

Researching this report

1. An electronic search of the scientific literature published during the previous 10 years, using the search terms 'antenatal testing', 'antenatal screening', 'prenatal testing', 'prenatal screening', 'genetic testing', 'learning disabilities', 'intellectual disabilities', 'learning difficulties', 'developmental disabilities', 'late termination of pregnancy', 'birth defects', and 'preimplantation genetic diagnosis'; the databases Medline, Cinahl, PUBMED, Cochrane and Psychinfo; and the Current Awareness Service published by the British Institute of Learning Disabilities. The abstracts of over 500 publications were examined.
2. Manual searches of relevant journals, including Journal of Intellectual Disability Research, Midwifery, Journal of Genetic Counseling, and Journal of Nursing Scholarship.
3. The search engine Google was used to search for relevant websites and literature (such as reports) not identified through other sources. Links to other websites were explored using a snowball technique. Websites were reviewed by the authors for relevance to families, professionals or both.
4. A letter (Appendix 1) sent electronically to national and international contacts of the authors giving details of the project and requesting relevant information about current or previous relevant research in the topic area. Key authors identified through the literature review were individually contacted by email for the same purpose.

A letter requesting information about current or previous relevant research in the topic area was posted onto the websites or circulated via the listserves of the following organisations:

British Society for Human Genetics
European Society for Human Genetics
International Society of Nurses in Genetics
Association of Genetic Nurses and Counsellors (UK)
National Society for Genetic Counselors (USA)
Australasian Society of Genetic Counsellors
International Association for Scientific Study of Intellectual Disabilities
Learning Disability Network (UK)
Learning Disability Network (Ireland).

Request for information sent out on relevant Listserves

Dear Colleagues

We are presently involved in undertaking a review of available information on antenatal screening and information provided for parents at the time of antenatal screening. This work has been requested by the Foundation for People with Learning Disabilities, which is a UK based charity providing information and research relating to services for people with learning disabilities (see www.learningdisabilities.org.uk).

The current project aims to explore issues for people with learning disabilities and their families arising from:

- antenatal genetic screening (and in vitro fertilisation embryo selection)
- late abortion of fetuses with disabilities
- genetic manipulation

The key objectives for this phase of the review are to

- find out what work has been done or is under way that is relevant to the project aims,
- what are the perspectives of different organisations active in this field
- and, in particular, what work has been done with people with learning disabilities and family carers on these issues.

If you are currently undertaking work in this area (or have previously done so) we would be grateful if you would share this information with us. We would also be grateful for copies of any information you have produced in relation to aspects of antenatal screening and people with learning disabilities.

Alternatively if the information is available on the internet, please provide the relevant website address. As this review is to be completed by the end of June, we would be grateful if you could respond to this request by 17th June 2005.

If you would like to discuss this review further please contact either of us at the addresses provided below.

We look forward to hearing from you.

Yours Sincerely

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APPENDIX II

Pathway to Down Syndrome Screening

Provided by National Down Syndrome Screening Programme

(Available at http://www.nelh.nhs.uk/screening/dssp/NSC_Flowchart_2.pdf, accessed 25th June 2005)

APPENDIX III Table of relevant websites

Host	Website address	Useful for: P=professional F=family
<p>Alix Henley website Excerpts from Pregnancy and Antenatal Care, Chapter 19 from Culture, religion and childbearing in a multiracial society.</p>	<p>www.alixhenley.co.uk/childbearingchapter.htm Accessed 17.7.06</p>	<p>P, F</p>
<p>AnSWeR Sections on Tests, Conditions, Choices, General, Disability, Ethics, Links</p>	<p>www.antenataltesting.info/ Accessed 17.7.06</p>	<p>F</p>
<p>Antenatal Screening Wales Screening tests Down Syndrome Downloadable pdf file</p>	<p>www.screeningservices.org/asw/public/screen_tests/screen_tests.asp www.screeningservices.org/asw/public/screen_tests/downs/downs.asp Screening for Down's syndrome in pregnancy (PDF file 572k) All accessed 17.7.06</p>	<p>Has dedicated sections for both P and F</p>
<p>ARC - Antenatal Results and Choices Making decisions Screening tests Diagnostic tests All antenatal tests Training for health professionals</p>	<p>www.arc-uk.org/ www.arc-uk.org/parent.html www.arc-uk.org/professionals.html Amended 17.7.06</p>	<p>F F P</p>
<p>Bart's and the London Hospital Antenatal Screening service What are DS, NTD and Edwards syndrome Downloadable information leaflets on screening Information on screening for professionals</p>	<p>www.wolfson.qmul.ac.uk/epm/screening/index.shtml www.wolfson.qmul.ac.uk/epm/screening/dsntd.shtml#top www.wolfson.qmul.ac.uk/epm/screening/leaflets.shtml www.wolfson.qmul.ac.uk/epm/screening/info.pro.shtml Accessed/amended 17.7.06</p>	<p>F F F P</p>

Table of relevant websites

Host	Website address	Useful for: P=professional F=family
<p>BioMedical Central BMC Medical Ethics Downloadable article on 'Antenatal screening and its possible meaning from unborn baby's perspective'</p>	<p>http://www.biomedcentral.com/1472-6939/2/3 Accessed 17.7.06</p>	<p>P</p>
<p>BioNews Weekly electronic newsletter – archive contains many articles on screening, testing</p>	<p>http://www.bionews.org.uk/ Accessed 17.7.06</p>	<p>F,P</p>
<p>Christian Medical Fellowship – excerpts from the Triple Helix The New Ethics of Abortion by John Wyatt Article on Late Termination by Jacky Engel</p>	<p>www.cmf.org.uk/literature/content.asp?context=article&id=886 www.cmf.org.uk/literature/content.asp?context=article&id=1086 Accessed 17.7.06</p>	<p>P P</p>
<p>Contact a Family Information on screening Contact a Family website has information on a large number of specific conditions that could be associated with learning difficulties. Examples given below: Fragile X syndrome DS Tuberous sclerosis Rett syndrome The organisation also provides factsheets and support for families.</p>	<p>www.cafamily.org.uk/screening.html www.cafamily.org.uk/Direct/f33.html www.cafamily.org.uk/Direct/d30.html www.cafamily.org.uk/Direct/t45.html www.cafamily.org.uk/Direct/r30.html All accessed 17.7.06</p>	<p>F,P F,P F,P F,P F,P</p>

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<p>Council of Europe Council of Europe, Committee of Ministers, Recommendation No. R (90) 13 on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling (June 21, 1990)</p>	<p>www1.urmn.edu/humanrts/instree/coerecr90-13.html Accessed 17.7.06</p>	<p>P</p>
<p>Department of Health Preimplantation Genetic Diagnosis (PGD) – Guiding Principles for Commissioners of NHS services</p>	<p>www.dh.gov.uk/PublicationsAndStatistics/Publications/PublicationsPolicyAndGuidance/PublicationsPolicyAndGuidanceArticle/fs/en?CONTENT_ID=4118934&chk=Dp24oj Amended 17.7.06</p>	<p>P,F</p>
<p>DIPEX General information about antenatal screening and diagnosis Questions and answers about screening Antenatal screening- parents experiences</p>	<p>www.dipex.org/EXEC Follow links to Q and A Follow links to parent experiences. Amended 17.7.06</p>	<p>F,P F,P F,P</p>
<p>Disabled Parents Network Offers support to disabled parents, no specific antenatal screening or testing info but would support parents making decisions if contacted.</p>	<p>www.disabledparentsnetwork.org.uk/about/aims.htm Accessed 17.7.06</p>	<p>F</p>
<p>Down's Syndrome Association Position statement on antenatal screening</p>	<p>www.dsa-uk.com/ www.dsa-uk.com/DSA_detStatement.aspx?st=11 Accessed 17.7.06</p>	<p>F,P F,P</p>

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<p>Down's Syndrome Scotland People with Down's syndrome Number of very good fact sheets on communication with families about Down Syndrome Information on screening and testing for DS Screening in pregnancy Information sheet for families – downloadable</p>	<p>www.dsscotland.org.uk/ www.dsscotland.org.uk/people-with-downs-syndrome/ www.dsscotland.org.uk/publications/professionals-students/ www.dsscotland.org.uk/news-and-events/press-releases/screening-and-testing www.dsscotland.org.uk/health/pregnancy/publications/families-friends-carers/screening-and-testing.pdf Accessed 17.7.06</p>	<p>F, P F, P F, P P, F F, P</p>
<p>European Society for Human Genetics Draft document on The Interface between Medically Assisted Reproduction and Genetics: Technical, Social, Ethical and Legal Issues</p>	<p>www.eshg.org/BGDocuAfterSevilla030605etAnnexes.pdf Accessed 17.7.06</p>	<p>P</p>
<p>For Parents by Parents Tests during pregnancy Personal experiences and links to sites for information</p>	<p>www.forparentsbyparents.com/pregnancy_tests.html Accessed 17.7.06</p>	<p>F</p>
<p>Forum for Science, Industry and Business for the advancement of innovation dynamics and the networking of innovation and performance potential Report on antenatal screening</p>	<p>www.innovations-report.de/html/berichte/medizin_gesundheit/bericht-18869.html Accessed 17.7.06</p>	<p>P, F</p>
<p>Genesense Interactive Educational Website Case study based information on antenatal screening and genetic testing for health professionals</p>	<p>www.genesense.org.uk Accessed 17.7.06</p>	<p>P, F</p>

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<p>Genomics Policy Unit (GPU) Telling stories, understanding real-life genetics</p>	<p>www.glam.ac.uk/socsschool/research/gpu/intro.doc Amended 17.7.06</p>	<p>P</p>
<p>Guild of Catholic Doctors Submission to Human Genetics Commission on antenatal screening Refusal form for antenatal screening tests</p>	<p>www.catholicdoctors.org.uk/By_Topic/TopicFrame.htm www.catholicdoctors.org.uk/Submissions/refusal_form_for_antenatal_tests.htm</p>	<p>F, P F, P</p>
<p>Health Promotion Agency The Pregnancy Book Includes information on antenatal screening. Book downloadable and relevant across UK</p>	<p>www.healthpromotionagency.org.uk/Resources/children/pregnancybook.htm Accessed 17.7.06</p>	<p>F</p>
<p>Health Technology Assessment Summary report on screening for Down syndrome Screening for fragile X syndrome: a literature review and modelling study by Song et al (downloadable)</p>	<p>www.ncchta.org/execsumm/summ2011.htm http://www.ncchta.org/execsumm/summ716.htm Accessed 17.7.06</p>	<p>P P</p>
<p>Health Technology Assessment A systematic review of antenatal screening</p>	<p>www.hta.nhsweb.nhs.uk/fullmono/mon711.pdf Accessed 17.7.06</p>	<p>P</p>
<p>Human Genetics Society of Australasia Best practice guidelines for offering antenatal screening</p>	<p>hgsa.com.au.thecoolserver.com/images/UserFiles/Attachments/Best%20practice%20GuidelinesantenatalscreeningforDownsyndromeC-Obs4Mar2004.pdf Amended 17.7.06</p>	<p>P</p>

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<p>Kids Health Information on range of tests, does not just focus on DS</p>	<p>www.kidshealth.org/PageManager.jsp?dn=KidsHealth&lic=1&ps=107&cat_id=&article_set=21958 Accessed 17.7.06</p>	<p>F</p>
<p>Leeds Antenatal Screening Service Information on Down syndrome Information on screening for DS Choice of tests Other disorders that could cause learning difficulties</p>	<p>www.intellectualdisability.info/home/about.html www.leeds.ac.uk/lass/screening%20for%20Down's.htm www.leeds.ac.uk/lass/choice_of_tests.htm www.leeds.ac.uk/lass/other.htm All accessed 17.7.06</p>	<p>F, P F, P F, P F, P</p>
<p>Leeds University Learning about intellectual disabilities and health – a web-based learning resource for medical and health care students and practitioners. Section on antenatal screening tests Academic Unit of Psychiatry and Behavioural Sciences information on research project titled: Couple's experiences of antenatal screening</p>	<p>www.intellectualdisability.info/home/about.html www.leeds.ac.uk/lass www.leeds.ac.uk/medicine/psychiatry/research/downs.htm All accessed 17.7.06</p>	<p>P P F, P</p>
<p>London IDEAS Gene Knowledge Park Report on workshop on antenatal screening and primary care</p>	<p>www.londonideas.org/internet/events/antenatalworkshop.htm Accessed 17.7.06</p>	<p>P</p>

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<p>March of Dimes Range of factsheets available to download Information on amniocentesis Special topic pages birth defects and genetics include: Down Syndrome Fragile X syndrome general information on genetic counselling (very US oriented)</p>	<p>www.marchofdimes.com/home.asp www.marchofdimes.com/professionals/14332.asp www.marchofdimes.com/pnhec/159_520.asp www.marchofdimes.com/pnhec/4439_1214.asp www.marchofdimes.com/pnhec/4439_9266.asp www.marchofdimes.com/pnhec/4439_15008.asp All accessed 17.7.06</p>	<p>F, P</p>
<p>Medical Journal of Australia Editorial on antenatal screening</p>	<p>www.mja.com.au/public/issues/172_07_030400/oats/oats.html Accessed 17.7.06</p>	<p>P</p>
<p>MIDIRS Informed Choice for Health Professionals Informed choice leaflets on antenatal screening. Information sent by email, leaflet for parents and a separate leaflet for professionals.</p>	<p>www.infochoice.org/ic/ic.nsf/TheLeaflets?openform Accessed 17.7.06</p>	<p>F, P</p>
<p>National Perinatal Epidemiology Unit Studies on: Women's expectations and experiences of antenatal services in Northern Ireland Women's experience of antenatal screening</p>	<p>www.npeu.ox.ac.uk/npeu_home.php www.npeu.ox.ac.uk/inequalities/index.php?content=inequalities_ni_study.inc http://www.npeu.ox.ac.uk/inequalities/index.php?content=inequalities_screening_study.inc All accessed 17.7.06</p>	<p>P P</p>

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<p>NELH Antenatal Screening - working standards incorporating those for the National Down's Syndrome Screening programme for England. Antenatal screening for Down's Syndrome - policy and quality issues</p>	<p>http://libraries.nelh.nhs.uk/screening/viewResource.asp?categoryID=1328&uri=http%3A//libraries.nelh.nhs.uk/common/resources/%3Fid%3D33974 http://libraries.nelh.nhs.uk/screening/viewResource.asp?categoryID=1328&uri=http%3A//libraries.nelh.nhs.uk/common/resources/%3Fid%3D60285 Accessed 17.7.06</p>	<p>P P</p>
<p>New Zealand, The National Screening Unit Screening for DS in New Zealand</p>	<p>http://www.moh.govt.nz/moh.nsf/wpg_index/About-National+Screening+Unit+-+down+syndrome#1 Accessed 17.7.06</p>	<p>P</p>
<p>NHS Direct Online Encyclopaedia Antenatal screening Topics: Introduction, When it should be done, Why it is necessary, How does it work, Results, What it is used for, Who can use it? Links.</p>	<p>www.nhsdirect.nhs.uk/articles/article.aspx?articleId=666 Amended 17.7.06</p>	<p>F</p>
<p>Patient Plus Information for patients on antenatal screening tests</p>	<p>www.patient.co.uk/showdoc/40001757/ Accessed 17.7.06</p>	<p>F</p>
<p>Patient Plus UK Information on screening for Fragile X syndrome</p>	<p>www.patient.co.uk/showdoc/40024830/ Accessed 17.7.06</p>	<p>F</p>
<p>Prenatal Screening and Prenatal Diagnosis Canadian website with information of both screening and testing</p>	<p>www.ahsc.health.nb.ca/prenatalscreening/ Accessed 17.7.06</p>	<p>F</p>

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<p>Pro Choice Forum Text of paper on Information giving and decision making in ante-natal screening By Joanie Dimivicius</p>	<p>www.prochoiceforum.org.uk/and2.asp Accessed 17.7.06</p>	<p>F, P</p>
<p>Public Health Genetics Unit Information on conditions Fragile X</p>	<p>www.phgu.org.uk/info_database/diseases/fragile_x/fragilex.html Accessed 17.7.06</p>	<p>P</p>
<p>Research project Moral considerations of pregnant women who are offered prenatal screening for congenital defects and the implications for policy-making. Netherlands 2002-2006</p>	<p>www.onderzoekinformatie.nl/en/oi/nod/onderzoek/OND1304818/ Amended 17.7.06</p>	
<p>Royal College of Obstetricians and Gynaecologists A Consideration of the Law and Ethics in Relation to Late Termination for Fetal Abnormality Report of the RCOG Ethics Committee</p>	<p>www.rcog.org.uk/index.asp?PageID=73&BookCategoryID=2&BookTypeID=4&BookDetailsID=329 Accessed 17.7.06</p>	<p>P</p>
<p>Royal College of Obstetricians and Gynaecologists Downloadable file on screening for DS (standards for practice)</p>	<p>www.rcog.org.uk/resources/public/pdf/down_syndrome_screening.pdf Amended 17.7.06</p>	<p>P</p>
<p>The Bub Hub (Australian online directory of pregnancy, baby and toddler services) Information for parents on screening and testing</p>	<p>www.bubhub.com.au/newsletterfeb0502.shtml Accessed 17.7.06</p>	<p>F</p>

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<p>UK National Screening Committee Antenatal screening subgroup Information on all aspects of the Down's screening program.</p>	<p>www.nsc.nhs.uk/antenatal_screen/antenatal_screen_ind.htm www.screening.nhs.uk/downs/home.htm Amended 17.7.06</p>	<p>P</p>
<p>University of Cambridge Centre for Family Research Genetics Group Research projects include: Genetic diagnosis and X-linked learning disorders (Helen Statham, Martin Richards, Maggie Ponder, Nina Hollowell and Lucy Raymond)</p>	<p>www.sps.cam.ac.uk/CFR/cfr_research_activity.htm Accessed 17.7.06</p>	<p>P</p>
<p>World Health Organisation Antenatal Screening And Birth Defects Surveillance</p>	<p>www.afro.who.int/drh/antenatal.html Accessed 17.7.06</p>	<p>P, F</p>

Foundation for People with Learning Disabilities

About the Foundation for People with Learning Disabilities

We promote the rights, quality of life and opportunities of people with learning disabilities and their families. We do this by working with people with learning disabilities, their families and those who support them to:

- do research and develop projects that promote social inclusion and citizenship
- support local communities and services to include people with learning disabilities
- make practical improvements in services for people with learning disabilities
- spread knowledge and information.

If you would like to find out more about our work, please contact us.

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The Foundation for People with Learning Disabilities is part of the Mental Health Foundation
Registered charity number 801130

ISBN: 978-1-903645-88-8

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