JUST PRENATAL TESTING?
The science, ethics, and policy of testing for Down syndrome

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Abstract

Prenatal testing for Down syndrome (DS) has been available for over forty years. With the development of screening technologies, testing is now offered to all pregnant women. But is the increasing use of these tests really a good thing? Is a test for DS just another test; or is it another form of disability prevention, one with unsettling and discriminatory overtones?

In this thesis I argue that whether testing is permissible or not is dependent in part on the morality of abortion. After an examination of arguments regarding fetal viability, sentience, and potential, I conclude that a fetus with DS may have less right to life than a fetus without DS, but this is dependent upon one’s perception of what constitutes ‘the good life’. That raising a child with DS may result in significant disruption to parents’ lives is an additional factor to take into account. If we hold that an abortion for social reasons is an acceptable expression of reproductive autonomy, then abortion for DS should be viewed as a morally permissible act.

While prenatal testing for congenital conditions is distinct from many routine pregnancy tests, it does not follow that testing for DS should be restricted. It is conceded that prenatal testing may offend some people with DS (and their families), on the ground that DS may have considerable bearing on the development of one’s identity. But while some people take offence at the prospect of testing and abortion, it is not clear that they should; for the decision to terminate a pregnancy is a complex one, based upon many different factors.

As new technologies become available, it is likely that more women will choose prenatal tests. More testing may mean that the numbers of those with DS will fall. However, this possibility should not preclude women access to testing. Pregnant women do not have a responsibility to ensure the continued prevalence of any trait or disorder, and DS is no exception.

The treatment of people with disabilities has improved over recent decades, and this is something one would expect to continue. Though the number of people with DS may fall, there is increasing likelihood that social support will be more comprehensive than in the past. Ensuring that this occurs is of utmost importance.
By using the controversial case of testing for DS, important issues surrounding all forms of testing in pregnancy come to light. This thesis concludes that two criteria must be met for a condition to be suitable for prenatal testing: that the condition significantly disrupts parents’ lives, and that the condition significantly limits a child’s open future.

Although the existing framework may need improvement, the current practice of prenatal testing enhances reproductive autonomy. It provides information on one’s pregnancy which many women find important, and enables decision-making which is otherwise unavailable. Though not without its challenges, prenatal testing for DS is morally defensible, and should continue.
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Introduction

Sarah Kay, eleven weeks pregnant, makes her second visit to see her midwife. Sarah is 23 years old, recently married, and is looking forward to the birth of her first child. The first visit with the midwife was two weeks ago, where she checked her tummy, gave Sarah some pamphlets, and made their next appointment.

This session is longer than the first one. As well as the usual tummy check and questions about how she is feeling, Sarah receives tests for rubella, syphilis and hepatitis B, along with several others that she can’t remember the names of. The midwife draws two vials of blood from Sarah’s arm, which she says will be sent off “to check your iron levels, and blood cells, and things like that”. At the end of the session, Sarah is told about screening for Down syndrome. The midwife informs Sarah that the test requires a blood sample like those that were taken before, and an ultrasound. The cost of the test is thirty dollars, but this only goes towards imaging. The blood test is free.

Sarah asks the midwife what she would do. The midwife tells Sarah that it is her choice and hers alone, but if she was in her position then she would take the test. She says that Down syndrome is associated with a much higher risk of miscarriage, and if born the child will have significant “mental problems”. Sarah was already looking forward to looking at her baby for the first time through ultrasound. As the blood test is free, she decides to have the test.

Later that week, Sarah is at coffee with three friends from work. Two of the women are much older than Sarah, and have families of their own. Her friends ask Sarah about her pregnancy. She tells them about her last visit to the midwife; about the multitude of tests, and the number of pamphlets she keeps getting given. She says that she is excited about her first ultrasound next week, but is a little worried as she hopes her baby is OK. When prompted, Sarah tells her friends that the test she is having is for Down syndrome.

This causes an argument at the table. Jane, who is 33, asks why Sarah feels that she needs the test. Sarah explains that it is only a blood test, and that her midwife thought that it was a good idea. Jane asks what Sarah would do if it turned out that her child did have Down syndrome. Sarah replies, “um... I’m not sure... I guess I would have to wait until I’m in that position...”. Jane is obviously angered at this reply, and she goes
on to explain that her twelve year old nephew has Down syndrome, and is “the sweetest little boy you have ever laid eyes on”. She thinks that it is ridiculous that you are able to test for something when the kids are so loveable, and that as a parent one should love a child regardless of “arbitrary medical conditions”. Jane urges Sarah not to have the test.

That afternoon, Sarah is scheduled to have her blood test. She thinks through all that was said earlier that morning at coffee. Jane’s description of her cousin sounded wonderful, and the family of which he was a part seemed incredibly supportive. Sarah wonders whether she would be able to do the same, if her child was born with the disorder. Though Sarah knows what people with Down syndrome look like, she has never met one, other than the girl three years below her at primary school; but that was years ago now. All the information she has on the disorder is the pamphlet that her midwife gave her. Sarah thinks back to how offended Jane was when she said that she was booked in for the test, and wonders what to do. She decides to call her husband.

In New Zealand around two hundred pregnancies are aborted due to the presence of fetal abnormality every year (Holloway et al., 2012). Fetal abnormality is detected by prenatal testing, such as the screening test for Down syndrome (DS) offered above. Currently, all pregnant women are offered prenatal screening, and over half accept. Of the 40,000 who receive screening, 1000 will be told that they are at increased risk of bearing a child with DS or other congenital condition (National Screening Unit, 2012). Most of these women will be referred for invasive testing to confirm or rule out the presence of fetal abnormality. If DS is confirmed, the chances are that the mother will abort the pregnancy.

Hence, cases like Sarah’s are not uncommon. Prenatal screening for DS and other conditions is now offered in most first-world hospitals. It is often presented within the first few meetings between a pregnant woman and her maternity carer, at a time when many other forms of testing are taking place. This is all within the context of varied recommendations to cease smoking, avoid certain foods and alcohol, and steer away from certain activities, to ensure the health of the developing child. Women may not be aware of what the screening entails, or what they will do if they receive a positive result.

Reactions like Jane’s are not uncommon either. Many feel strongly that prenatal testing for congenital disease is wrong, often because they believe that selecting on the basis of genetics is not being a responsible parent, or because they think that the
identification of disorders that may lead to abortion demeans those with disabilities. Testing for DS is especially divisive, as the condition is not directly associated with pain, suffering, or a major reduction in life expectancy. Indeed, some parents say that children with DS are 'happy kids', who are relatively easy to raise.

Tackling the question of whether it is right to test for DS is illuminating, as it may shed light on the morality of testing for other more life-affecting disorders. If it is ethically acceptable to test for DS, then it may be acceptable to test for these other conditions, as well.

The first part of this thesis examines the science surrounding DS and prenatal testing. Along with a brief history, Chapter 1 asks what it is like to live with DS, and what it is like to live in a family of someone with DS. Chapter 2 highlights the difference between screening and diagnosis, and asks what kind of prenatal tests lie over the horizon. These foundations are necessary if we are to approach the ethical problems raised by prenatal testing.

The second part of this thesis discusses the ethics of testing for DS. To do this, I ask two key questions. Chapter 3 asks if it is morally permissible to abort in the case of a fetus with DS. Chapter 4 asks whether the offer of prenatal testing for DS could be discriminatory in a way that other tests are not.

The third part of this thesis addresses the policy of prenatal testing. Chapter 5 studies whether the current practice of prenatal testing is ethically sound. Chapter 6 is a summary, suggesting notes for policy from the thesis as a whole.

Throughout the text, other questions will be raised. Among others, these include whether testing and abortion is consistent with the ideals of parenting, whether DS is an inseparable part of one's personal identity, and whether prenatal testing will eventually cause DS to vanish.

Many of my arguments raise significant issues for our approach towards disability. However, a particular strength of this thesis is that it avoids the confusion which surrounds the term ‘disability’, exposing the weakness of any arguments by confining them to a specific congenital disorder.
It is also of note that my thesis makes little reference to the partner's role in decision-making. This is simply a formal necessity; though women can and often do discuss the meaning, role, and choice of prenatal testing with their partners, not all do, as some women are single, or do not involve their partner in decision-making. To make my argument applicable to all women, I will often mention only the woman in the context of pregnancy care.

Lastly, I refer to ‘people with DS’, to emphasize the individual and not the disorder (as opposed to ‘DS people’). This is in line with most disability rights perspectives, which suggest that we should emphasize the person (e.g. ‘people with disabilities) as opposed to the disability (e.g. ‘disabled people’) (Support Advanced Learning and Training Opportunities, 2006).
Part One: Matters of science
Chapter 1  DOWN SYNDROME: AN OVERVIEW OF A GENETIC CONDITION

Down syndrome (DS) is an autosomal genetic anomaly which affects between one in 600 to one in 1000 live births (Sindoor, 1997). It is the most common chromosomal abnormality among newborn infants, and the most common cause of cognitive impairment. DS affects all cultures, and there does not seem to be any social, economic, gender, or race predilection.

1.1 History of Down syndrome

Although DS has probably been present throughout the development of the human race (Czarnetzki et al., 2003), it was not until 1838 that a patient with features characteristic of DS was recorded. This appeared in the first handbook of psychiatry, written by Jean-Etienne-Dominique Esquirol (1838). DS was noted under a section titled ‘Idiocy’, where a category of patient was characterized by a protruding tongue, epicanthic eye folds and flat nasal bridge. Malformed limbs, short stocky stature and ‘mental retardation’ [sic] were also mentioned (Roubertoux, 2006).

A similar classification was adopted in 1846 by Edouard Seguin, another French physician who worked with the cognitively impaired. He built on the description from Esquirol, adding that the tongue was thick and cracked, and the skin and lungs were especially susceptible to infection (Roubertoux, 2006). Seguin later noted that in spite of profound cognitive impairment, those with DS were “good kids” who “had language and were able to gain some basic knowledge” (Seguin, 1856, cited in Roubertoux, 2006, p. 346). Seguin labelled this class of conditions as ‘furfaraceous cretinism’, but this classification was not readily accepted. Indeed, the definition we use today has very different origins.

Johann Friedrich Blumenbach was a German physician and anthropologist. In 1779 he divided the human species into five races, a classification system which proved to be very influential. The terms were still in use a century later, shown by an article titled ‘Observations on an Ethnic Classification of Idiots’ published in ‘London Hospital Reports’ in 1862. The article gave congenital impairment a five class system, based on a condition’s similarity to each of Blumenbach’s main races (Caucasians, Aztecs, Malayans,
Ethiopians and Mongolians). The purpose of this was to enable a newborn diagnosis, allowing the physician to describe the current condition and future of the neonate. All five races were covered, but it was to the 'Mongols' that much of the article was devoted. The author's name was John Langdon Down (1866).

Today, Down (1866) is given most of the credit for the discovery of DS, although the disease was first noted two decades earlier. The disease came to be called 'mongolism', such was the influence of Down’s article, where he vividly describes the hallmark symptoms of DS:

The hair is not black, as in the real Mongol, but of a brownish colour, straight and scanty. The face is flat and broad, and destitute of prominence. The cheeks are roundish, and extended laterally. The eyes are obliquely placed, and the internal canthi more than normally distant from one another. The palpebral fissure is very narrow. The forehead is wrinkled transversely from the constant assistance which the levatores palpebrarum derive from the occipito frontalis muscle in the opening of the eyes. The lips are large and thick with transverse fissures. The tongue is long, thick, and is much roughened. The nose is small. The skin has a slight dirty yellowish tinge, and is deficient in elasticity, giving the appearance of being too large for the body. (Down, 1866, cited in Down, 1962, p. 417)

The way that Down describes the symptoms of DS would not be acceptable today, yet is typical of the language and the approach towards disability of the time. Down (1866) noted that the 'mongoloid' [sic] symptoms were reasonably prevalent, occurring in over 10% of cases of cognitive impairment. He recognised that the condition was a congenital disorder, although he wrongly believed that parental tuberculosis was the cause (Roubertoux, 2006).

Although the term 'mongolism' is now seen as racist and discriminatory, Down’s (1866) comparisons of the syndrome to the 'Mongol' people were meant to portray the exact opposite. At the time there were many who believed that separate human races had separate biological origins (a school of thought named polygenism). Down believed that his 'ethnic classifications' refuted this, as two parents of one race could produce
offspring that instead resembled that of another. Down suggested that DS supported the idea that all races were unified; and therefore, that none was superior over another.

The collection of the aforementioned traits began to be called ‘mongolism’, a term which was still in wide use nearly a century later (Penrose, 1961). In the 1960s the alternative definition of Down's Syndrome was suggested due to the distress the word caused to parents, together with mongolism's racial connotations (Spalding, 1961). Thereafter, efforts were made to phase out the use of ‘mongolism’ completely, though it was still used in the literature until the early 1990s (Leung & Seagram, 1991). Down’s Syndrome has since been altered to ‘Down syndrome’ in accordance with other syndrome descriptions (e.g. Guillian-Barre syndrome, Munchausen syndrome and Turner syndrome).

Trisomy 21 has been suggested as a more accurate term than Down syndrome. This is for two reasons; Esquirol, not Down, wrote the first records of those with DS; and it correctly describes the aetiology of the disease (Roubertoux, 2006). Despite this, ‘Down syndrome’ continues to be the term most widely used.

1.2 Aetiology of Down syndrome

The cause of DS was elucidated by Jerome Lejeune in 1958, when he found that a ‘mongoloid’ [sic] patient had 47 instead of 46 chromosomes (Lejeune, 1959). This was found to be present in nine other patients with DS (Lejeune et al., 1959), and later confirmed by three independent clinical groups.

It was first noted by Penrose (1951) that increasing maternal age was related to an increased chance of bearing a child with DS. This is now confirmed as being due to the increase in risk of non-disjunction in meiosis (Yoon et al., 1996). This non-disjunction leads to an unequal chromosome arrangement in oocytes, resulting in aneuploidy of fertilized embryos (Koehler et al., 1996; Roizen & Patterson, 2003). Non-disjunction pre-conception contributes to around 94% of cases of DS (Chen, 2012). As shown in Figure 1, the chance of giving birth to a child with DS climbs rapidly after age 35 due to the increased likelihood of non-disjunction.
Although more likely in meiosis, non-disjunction may also take place in the early development of the embryo. During the first few divisions of cells, a cell resulting from mitosis may be left with three 21st chromosomes. If this cell continues to divide, all resulting cellular offspring will exhibit trisomy 21 too. If this happens soon after conception, a significant part of the resulting fetus will have trisomy 21. This condition is termed ‘mosaic’ DS, and makes up around 2.4% of cases of DS (Chen, 2012).

The remaining 4% of DS cases are caused by translocation (Chen, 2012). Translocations describe when part of a chromosome is attached to another, usually separate chromosome. Together with mosaic DS, these conditions are probably not linked to maternal age (Caron, 1999). Regardless of the cause of trisomy 21, all people with DS have a significant degree of cognitive impairment, and the characteristic features noted by Down (1866) are shared by almost all.

Along with advanced maternal age, other risk factors for fetal abnormality include previous fetal abnormality and family history of aneuploidy, and environmental factors such as smoking and folic acid deficiency in the mother (Caron, 1999). While the link between advanced maternal age and DS has been recognised for many years, a link between paternal age and DS remains controversial (Sartorius & Nieschlag, 2010). One study suggested that the paternal contribution to DS is as high as 50% when the mother...
is aged over 40 (Fisch et al., 2003). However, the study found no paternal contribution when the father is aged 35 or less. Other large studies have found no significant association (De Souza et al., 2009; J. D. Erickson, 1978; Hook et al., 1995). If a link between DS and paternal age exists, it is likely that the real-life implications for this are small, compared to the relevance of maternal age (Sartorius & Nieschlag, 2010).

1.3 Clinical features of Down syndrome

Trisomy 21 can affect almost every organ system, yet some organs are affected more frequently than others. Because of this, people with DS are at high risk of many different congenital and acquired disorders.

DS is the leading cause of cognitive impairment, and all people with DS show some measure of intellectual disability (Roubertoux, 2006). Intellectual Quotient (IQ) ranges from 20-85, with the mean being approximately 50; 100 is the normal value of a standard population (Chen, 2012). Cognitive ability is usually in the moderately to severely impaired range, and mental age is rarely above eight years (Gibson, 1978). In simple tasks those with DS have normal performance, yet they lack ability with spatial, long term and procedural memory (Roubertoux, 2006). There is often a greater rate of decline in cognitive ability in adult life (Roubertoux, 2006).

Together with some degree of intellectual disability, similar phenotypic features are the only other traits which are shared by all people with DS. Children with DS are often described as having a happy disposition (Gilmore et al., 2003). Typical features of a child with DS such as the flat face, flat upper region of the nose and an open mouth are shown in Figure 2. Other phenotypic features of DS include:

- Shortness of the fingers and toes (brachydactyly)
- Broad hands
- Skin fold of the upper eyelid (epicanthal folds)
- Fifth finger bending towards other fingers (clinodactyly)
- Short stature
- Wide 1-2 toe gap
- Folded or small ears
- Abnormal teeth
Short neck
Excess skin at back of neck (Roizen & Patterson, 2003; Up To Date, 2012c)

Figure 2 Typical facial features of a child with Down syndrome. Image from Bickley (2009, p. 861)

People with DS are commonly described as being gentle, caring, and tolerant, as well as naturally spontaneous and patient (Roizen & Patterson, 2003; Sindoor, 1997). With all individuals there is variance, and some people with DS have frequent episodes of anxiety, can be stubborn, and strongly resist change (Chen, 2012; Sindoor, 1997).

Children with DS exhibit developmental delay, with developmental milestones taking around twice the time to be attained. For example, the average age of a child with DS to utter their first words is 18 months, as opposed to 10 for other children (Up To Date, 2012c). Seventeen per cent of children with DS have a psychiatric disorder; by adulthood, this number has risen to 25% (Chen, 2012). Aggressive behaviour forms a significant proportion of these disorders in both age groups. Attention deficit hyperactivity disorder (ADHD) can be common in children, and major depressive disorder may be common in adults. Autism has been linked with DS, and the frequency of this dual diagnosis may be as high as 7% (Kent, 1999).
People with DS typically have a short stature, and exhibit poor muscle strength which improves with age (Chen, 2012). Frequently, they have problems with speech. Features typical of ageing occur prematurely in those with DS, resulting in a loss of hair, decrease in skin tone and increased risk of senile dementia (Hasle, 2000). Some loss of hearing is common in people with DS as they age (Bittles & Glasson, 2004).

Those with DS are at much greater risk of a large number of congenital defects. The vast majority of these are not life-threatening, yet they can be debilitating and may lead to further illness. The most common abnormality is congenital heart disease, occurring in just under half (45%) of all patients (Freeman, 1998). This is the most significant factor determining life expectancy in individuals (Chen, 2012). In addition to cardiac problems, there is a high prevalence at birth of gastrointestinal, immunological, respiratory, sensory, and orthopaedic problems (Bittles & Glasson, 2004).

As well as being at increased risk of congenital malformations, those with DS are at higher risk of developing morbidity during life. People with DS are immunocompromised, and are at 12 times the risk of developing infectious disease (Chen, 2012). Children have a significantly increased risk of developing leukaemia. Thyroid disorders are common, with 40% of individuals developing a disorder by adulthood (Dinani & Carpenter, 1990). Adult onset epilepsy affects nearly half of those with DS over 50 years of age, becoming common after 30 years (Bittles & Glasson, 2004). Most of those with DS share neuropathological features common to Alzheimer’s disease by the age of 40, and dementia is diagnosed in at least 56% of those aged 60 or above (Bittles & Glasson, 2004). Other significant health problems found in adults with DS include acquired cardiac disease, pulmonary hypertension, chronic respiratory changes and loss of vision.

Fertility is affected in DS to varying degrees, and is dependent on gender. Females are able to have children, though they have markedly reduced fertility, and males are almost always infertile (Roizen & Patterson, 2003). Though children have arisen from both genders (Corker et al., 2011), relatively few offspring have been born with fathers who have DS (Bittles & Glasson, 2004).

Life expectancy for those with DS has increased dramatically throughout the last century. Evidence collated by Bittles and Glasson (2004) suggests that life expectancy for a child with DS was nine years of age in 1929, increasing to 12 years two decades
later. Life expectancy began to increase more rapidly in the 1950s, a trend which has continued to this day.

As shown in Figure 3, the gain in life expectancy of DS is well in excess of that of the general population, suggesting that people with DS will approximate normal life expectancy in the near future (Bittles & Glasson, 2004). This marked increase is related to increased antibiotic use and de-institutionalization. This resulted in reduced infection rates, which historically were a significant contributor to mortality.

![Figure 3 Global trends in life expectancy estimates for people with Down syndrome compared with those for the general Australian population, 1900-2000. Image from Bittles & Glasson (2004, p. 283)](image)

There is currently no cure for DS, and although research continues no viable treatment exists (Dierssen, 2012). Instead, interventions are aimed at the many structural deformities and co-morbidities associated with the disease. The most common of these is surgical correction of congenital cardiac defects, which has further contributed to the dramatic increase in life expectancy (Bittles & Glasson, 2004).
1.4 Living with impairment: Down syndrome and disability

People with DS exhibit similar phenotypic features, have some degree of cognitive impairment, and almost always live with several co-morbidities. Together, these form limitations, acting to prevent some processes, functions or actions. How, then, do these limitations constitute a disability? Indeed what is disability, and what does it mean to be disabled?

1.4.1 Defining disability

Persons with disabilities include those who have long-term physical, mental, intellectual or sensory impairments which in interaction with various barriers may hinder their full and effective participation in society on an equal basis with others. (United Nations Convention on the Rights of Persons with Disabilities, 2006, Article 1)

This is one of many definitions which have been suggested to explain 'disability'. It is useful because it differentiates between what are impairments (the actual functional deficit of a state or condition) and what is disability (the often negative experience of living with impairment). Thus, when we discuss disability we discuss the holistic impact of a functional deficit on an individual's life. In other words, we examine the interaction between a health condition and the environment (World Health Organisation, 2011).

This strikes a middle ground between the two major approaches towards disability. The oldest (and arguably the most influential) approach is now labelled the 'medical model', which suggests that the character of a person's impairment was the only factor which decided the experience of living with a condition. The 'medical model' is helpful as it indicates a way to improve one's life; through medical intervention to reduce the severity of the impairment. Total reliance on the medical model is unhelpful, however, as many impairments cannot be treated, let alone cured.

How, then, can lives with incurable illness be made better? Here an opposing school of thought is helpful. The 'social model' states that it is the social environment which is the only factor which decides the experience of living with a condition. Therefore the way to improve this experience is to minimize the social attitudes of stigma and discrimination against those with disability. It is not only social interaction
that can be debilitating, but the physical environment can be limiting, too. Examples of ‘cures’ for the social environment might include special education programmes for the cognitively impaired, or easy-access ramps for those with wheelchairs.

But the social model is also limited, as it disregards the importance of the medical condition for a person’s life. The patient who is blind will never be able to see, no matter how well she is integrated into society. The patient with multiple sclerosis will still suffer pain, though wheelchair access is provided throughout her city. As a result, the experience of impairment will still be limiting, and at least a little negative. As Adrienne Asch (1989b) writes, “the inability to move without mechanical aid, to see, to hear, or to learn is not inherently neutral. Disability itself limits some options” (p. 73). The social model regards the impairment as ‘value neutral’ for the experience of disability, and thus complete reliance on the social model is inadequate.

What is more appropriate is a bio-psycho-social approach, one which understands that an individual’s experience of living with a condition is a product of both the impairment, and the social constructs and ways of thinking of disability (Shakespeare, 2006; World Health Organisation, 2011). This provides us with several ways to reduce the negative aspect of living with impairment; by acting to reduce the impairment through medical intervention, acting to remove discrimination with legislation, and social support and extra provisions for the disabled.

With regards to DS, the syndrome cannot be cured. Though there are several avenues for treatment, any efficacious therapy looks beyond the near future in humans (Dierssen, 2012). Medical intervention is limited to treatment of the co-morbidities associated with DS. Social interventions, therefore, are probably the best course of action to mitigate the negative aspects of living with DS.

1.4.2 What does it mean to have Down syndrome?

Though disabilities all share some form of limitation, in reality all disabilities are different. As disability is a lived experience, the same impairment for two people may affect them in very different ways. Whereas one may feel that a disability does not prevent her from doing what she finds special, another may find that her life is consistently thwarted and dreams are dashed (Shakespeare, 2006).
Therefore it is hard to generalize what DS means to a specific person with DS, a task made even harder by the wide spectrum in the severity of the disease. Whereas one individual may have only mild cognitive impairment and be moderately independent, another may be severely impaired, heavily dependent on others and have many co-morbidities. Though both fall under the umbrella of ‘DS’, in effect their lives are very different. However, some attempts can be made to generalize about the lived experience of the disorder, and how this might compare to those who do not have DS.

Individuals with DS have varying levels of ‘self-concept’; that is, how one thinks and feels about oneself. In development these have been separated into distinct stages: self-recognition, self-representation, self-description, self-assertion, self-regulation and self-evaluation (Skotko et al., 2011c). These stages are common to both DS and non-DS development, though progression through the stages for those with DS is delayed (Skotko et al., 2011c). This suggests that people with DS have similar ways of thinking about their lives as people without DS.

As noted by Seguin in 1848, those with DS are frequently described as friendly, happy and affectionate. However, this stereotype is not always true. Another stereotype entrenched in medical literature is the idea that those with learning difficulties have lower self-evaluations of themselves, comparing their limitations negatively with others’ abilities (Montgomery, 1994). Again, this is inaccurate, especially when applied to DS. Though research on this subject is limited, it has progressed in the last two decades. Research by Amanda Begley (1999) indicated that similar to that of younger children without learning difficulties, “pupils with DS hold very positive self-perceptions [of themselves]” (p. 515). This study corroborated findings from a smaller study several years earlier (Cuskelly, 1996).

Young adults with DS have also been assessed on their self-evaluation. Those with DS consistently rate themselves positively on measures of self-esteem (Cunningham, 2004; Glenn, 2001), with a strong bias towards positive comparisons (Glenn & Cunningham, 2004). Positive comparisons are ‘upward’ comparisons (i.e. “I’m better than”) compared to ‘downward’ comparisons (i.e. “I’m worse than”).

The self-esteem of adults with DS has not been comprehensively assessed in academic literature. The largest study to date was a questionnaire study developed by Skotko et al. (2011c) and carried out in the United States (US). Skotko et al. received
responses from 284 people with DS aged 12 and older, who answered questions on their self-esteem. Their responses were overwhelmingly positive, indicating that “the overwhelming majority of people with DS are happy with their lives... who they are and how they look... [and] love their families, including their brothers and sisters” (p. 2368). Furthermore, the “majority of people with DS feel they can easily make friends... [and] feel that they help other people” (p. 2368). Perhaps unsurprisingly, only a small percentage of those who responded felt sad with their lives.

Though the study was large, it had several limitations. The response rate was low at only 15%, and the questionnaire is therefore at high risk of non-response bias (Skotko et al., 2011c). Those with DS with significant limitations may not have understood the questions, and if a large number of these felt that their impairments were negative this may have given an overly positive picture. Indeed, some who felt that their lives were harder to live may not have returned the questionnaire.

In addition, the average income of respondents’ families was twice that of the US median, meaning that the responses are not necessarily a representative sample, both for people with DS in the US and around the world. However, the data from such a large number of participants is helpful in assessing the self-evaluations of people with DS; one which seems to be, overall, very positive. Though Skotko et al. (2011c) did not compare these perceptions to those of the non-DS population, one could surmise that these evaluations would be similar, if not more negative, than their DS counterparts.

Evidence on self-evaluation of people with DS is limited. But what has been gathered indicates that those with DS are happy with their families, their friends, and their abilities. More evidence is needed to supplement these findings, preferably from studies with higher response rates. However, it seems more than reasonable to conclude that people with DS are not hugely troubled by their disability, and are happy with themselves and their lives.

1.5 Down syndrome and the family

As any parent will be aware, all children require significant input in time, money, and resources for their upbringing. Children with DS are no different. Indeed, due to their added limitations and extra needs, they often require greater support from parents and family. This is sometimes described as the added ‘burden’ of raising a child with DS;
for all children can be burdensome, but those with DS require more care than other children (Gath, 1990). For the purposes of this thesis I will avoid the usage of the value-laden term ‘burden’, as it can suggest that increased care is undesirable, and raising a child a chore- which is certainly not true for many parents.

Nevertheless, the question remains: do children with DS require more care than other children, or other children with disabilities? This is a problematic area of research, as DS presents with varying levels of intellectual ability, independence and co-morbidity. Furthermore, studies on more abstract values such as ‘love’ and ‘value’ are subjective in their nature. However, research has endeavoured to elucidate the value of children with DS’s lives for both the parents of those with DS, and their siblings. It is to this literature that we now turn.

1.5.1 Outcomes and perspectives of parents

Where children with DS are compared to those with other disabilities, the literature has indicated a ‘DS advantage’ in many areas (Cuskelly et al., 2008). The advantage has been repeated in many studies, detailing less cases of depression, children being easier to raise, marriages and families being happier and closer, and parents reporting more positive experiences (Cuskelly et al., 2008).

Several explanations have been given for this ‘advantage’. The first is that children with DS are, themselves, less disruptive to the family unit. However, the DS advantage may have been due to variable income among groups in many of the studies, since when income was accounted for in the advantage disappeared (Stoneman, 2007). Another explanation is that DS families typically have older parents, and these marriages may be more stable (Corrice & Glidden, 2009; Skotko et al., 2011b). Though conclusive evidence is yet to appear, it does seem that raising a child with DS may be less disruptive than raising children with other disabilities.

Children with DS have also been compared to children without DS. There are some advantages; parents are less likely to get divorced (Urbano et al., 2007), and children are reportedly easier to raise (Marcovitch et al., 1987). Again, it is of note that both of these outcomes could be due to later marriages being more stable.

Some qualities are similar between families, regardless of having a child with DS: these include similar levels of marital satisfaction, levels of stress, cohesiveness, and
self-efficacy in parenting skills (Skotko et al., 2011b). There are some significant disadvantages that families that have a child with DS may experience: the children have more behavioural problems (Gau et al., 2008; Stores et al., 2002), some parents experience greater stress, depression and anxiety (Gau et al., 2008; Roach et al., 1999), and more time is spent on caregiving activities (M. Erickson & Upshur, 1989).

The last point is an important one. Children with DS are more dependent than children without DS, at all stages of development. Parents of children with DS have more caregiving difficulties and child-related stress (Roach et al., 1999). Not only do they devote more time to childcare, but spend less time on social activities (Barnett & Boyce, 1995). In most circumstances, an adult with DS never becomes fully independent. As those with DS have individual, special needs this may not seem surprising. But it is important to note, as children always require a large time investment. In the case of DS there is significantly increased time needed for caregiving, which in many cases is required throughout life. For some parents, this will be undesirable.

It is important to identify common perspectives of parents of children with DS, to see how they feel about the limitations and added needs of their child. This may indicate whether parents feel that raising their child with DS is a challenge, or no different to raising other children.

Studies of this kind are rare, but one was recently undertaken by Skotko et al. (2011b). Nearly 1500 families participated in the study, with the vast majority of respondents agreeing that they love and are proud of their child. A small minority (1 in 20) reported that they were embarrassed, and/or regretted having a child with DS. Other important findings were that parents felt their outlook on life was more positive because of their child with DS, and that their other children have good relationships with their child with DS, and are more caring and sensitive as a result.

Again, this study was subject to significant non-respondent bias due to a response rate of 29% (Skotko et al., 2011b). However, that such a large study showed very positive results is significant, indicating that the majority of parents of children with DS are proud of their child.
1.5.2 Outcomes and perspectives of siblings

Parents are not the only members of a family, and the outcomes on siblings of children with DS are also of note. Siblings of children with DS have no more behavioural problems than siblings of children without DS (Cuskelly, 1998). There is also no difference in social competence between these groups (Cuskelly et al., 2008; Van Riper, 2000). These siblings have similar self-worth when compared to siblings of non-DS children (Cuskelly et al., 2008). For the most part, siblings of children with DS take on additional responsibilities of care independent of gender, which they see as a positive interaction and not a burden [sic] (Cuskelly, 2003).

Having a brother or sister with DS has also been shown to have benefits. Siblings of those with DS show more empathy and kindness towards their brothers and sisters (Cuskelly, 2003), and have more warmth and less conflict in their relationship (Fisman, 2000). The ‘DS advantage’ also extends to their siblings; the sibling relationship is more positive and respectful than relationships with a child with autism (Knott et al., 2006).

Aside from the outcomes for siblings, the perspectives of siblings are also important. Skotko et al. (2011a) found that almost all siblings express love and pride for their siblings with DS, and this is regardless of age. This love is not dependent on the functional or health status of the sibling with DS. In addition, many children feel that their lives are enriched because of their siblings with DS, are happy assuming increased responsibilities, and plan to be involved with their sibling throughout life. Almost all siblings feel that they have a good relationship with their brother or sister with DS, and would not trade their sibling for another who did not have DS. Though some children were embarrassed by their sibling with DS, this was not true of the vast majority.

Similar to Skotko et al’s (2011a) aforementioned studies, the low response rate (19%) may mean that non-respondent bias is a factor in these findings. However, that the views of 822 children were so unanimously positive is encouraging. It indicates that for almost all siblings, the experience of having a brother or sister with DS is a happy one.
1.6 Conclusion

DS is the most common congenital abnormality at birth. The characteristic features of DS have been noted for some time, features that were once labelled ‘mongoloid’ [sic]. Despite being similar in some ways, people with DS are more different than they are alike. These outcomes cannot be predicted by current prenatal testing. At one time unlikely to live to their teens, those with DS now have a life expectancy of around 60 years. Due to improvements in care and awareness of related disease, life expectancy is expected to continue to improve.

From the above evidence, several points are clear. Those with DS are almost always valued by their parents, siblings and themselves. People with DS form an integral part of many families, and often allow siblings and parents to learn more about themselves- in terms of patience, love and ability. However, raising a child with DS is not without its challenges. Children with DS require significantly more care than other children, particularly when other behavioural disorders are present. These children will require special education, greater levels of care, and more support throughout life. However, the impairment of DS does not result in unhappy, unmotivated children. On the contrary, people with DS tend to be positive in their thinking and are socially active, having fulfilling relationships with family and peers.

Deriving these relationships is difficult and beset with challenges. That DS results in a range of intellectual disability means that some questions will be comprehended by some and not others, excluding the most cognitively impaired from studies. In addition, sample sizes have either tended to be too small to detect a significant relationship, or large with low response rates leading to selection bias. Nevertheless, the number and breadth of studies that have found similar relationships indicates that the experience of parenting a child with DS is a positive one. Though raising a child with DS may have significant demands, so does the raising of any child; as shown by the literature, this experience need not be a negative one.

However, it is reasonable to assume that although raising a child with DS is one which many embrace, it is also one which many parents will want to avoid. It is for this reason that prenatal testing is offered to pregnant women. But what methods of prenatal detection exist? And are some more or less problematic than others?
Chapter 2 Prenatal Testing: Scientific and Clinical Issues

The discovery in 1958 by Jerome Lejeune that DS had a genetic aetiology was ground-breaking. Never before had intellectual disability been directly linked to a specific chromosomal abnormality. DS was identified to be caused by three copies of the 21st chromosome, henceforth known as ‘trisomy 21’. The finding is all the more astounding given that the chromosome number of *Homo sapiens* was elucidated only two years prior (Tjio, 1956).

The detection of trisomy 21 led to further research into gene based, disease causing malformations. Lejeune himself proceeded to identify the genetic basis of Cri du Chat syndrome, 18q- syndrome, and other congenital disorders. To this day, trisomy 21 remains the most widely studied model of human aneuploidy.

The elucidation of the genetic basis of DS indicated the association between genes and congenital disease, but did not allow clinicians avenues for treatment or prevention. But with the increase in availability of karyotyping and the development of amniocentesis, this was about to change.

### 2.1 Prenatal diagnosis for Down syndrome

Amniocentesis is a procedure which involves the use of a needle to remove a small amount of fluid from the amniotic sac, as shown in Figure 4 (Simpson et al., 2012). The amniotic sac surrounds the fetus, containing amniotic fluid. The tapping of this fluid had been practiced for over a century before its recent widespread use (Woo, n.d.). Amniocentesis had previously been used to treat hydraminos (excessive amniotic fluid), and to test for Rhesus blood type incompatibility between mother and child (Cowan, 1994). It is now used for the prenatal diagnosis of genetic disease.

Diagnosis using amniocentesis was achieved in 1956 by two Swedish researchers, Fuchs and Riis (1956). This was the first example of prenatal diagnosis, and was important for those who had sex-linked disorders who wanted to avoid having a child with a particular (sex-linked) disease. From the 1970s amniocentesis began to be utilised in high numbers in prenatal care around the world (Cowan, 1994). It is now
supplemented with ultrasound to help guide the needle and prevent damage to mother or fetus.

![Figure 4 The procedure of amniocentesis, showing a needle extracting amniotic fluid from the amniotic sac under ultrasound guidance. Image from www.uptodate.com](image)

Best practice guidelines developed by the Royal College of Obstetricians and Gynaecologists (2010) state amniocentesis should be carried out at 15 weeks of pregnancy. Amniocentesis before this time has a higher fetal loss rate and risk of morbidity (Royal College of Obstetricians and Gynaecologists, 2010). Because of this, chorionic villus sampling (CVS) is offered at 14 weeks gestation or earlier (National Screening Unit, 2009).

CVS is a similar procedure to amniocentesis, which instead of utilizing amniotic fluid obtains part of the chorionic villi (the fetal blood vessels of the placenta). Two approaches are possible; via the cervix (transcervical), or through the abdomen (transabdominal), shown left and right respectively in Figure 5. Attempts were made to develop this technique in the late 1960s to allow earlier elective abortions for women (Cowan, 1994). Due to high rates of spontaneous miscarriage associated with the procedure, it has not been until recent years that CVS has had a place in standard care.
Both amniocentesis and CVS function as prenatal diagnostic techniques. Each procedure harvests cells viable for karyotyping. A ‘karyotype’ describes the number and appearance of chromosomes from a cell, as shown in Figure 6. It was first found in 1966 by Mark Steele that amniotic fluid cells are suitable for karyotyping, as the fluid contains cells which have separated from the fetus. Two years later, the first prenatal diagnosis of DS was reported by Henry Nadler (1968).

Karyotyping remains the primary method used to identify chromosomal abnormalities that can be detected under the microscope (Newberger, 2000). Karyotyping detects all forms of DS (non-disjunction, mosaicism and translocation) if present in the cell sample. It is of note that as CVS samples only the placenta it may not detect mosaicism. Because of this, some concern remains over the diagnostic use of CVS, though the prevalence of mosaic DS is low (Alfirevic et al., 2009).
Currently, test results from amniocentesis or CVS take around seven to 10 days to become available, as cells must be stimulated to grow (Up To Date, 2012a). The most common abnormality detected by amniocentesis is DS, followed by sex chromosome abnormalities and trisomy 18 (Caron, 1999). Other aneuploidies, such as trisomy 13, and structural rearrangements, such as achondroplasia, can also be detected (Wilson, 2000). Nowadays amniocentesis is used almost solely for detecting fetal abnormality, though it may still be indicated for excess amniotic fluid. It is important to note that no testing for DS can predict the severity of cognitive impairment, likelihood of comorbidity, life expectancy, or independence of the future child’s life.

Diagnostic procedures have one main aim: to confirm or rule out the presence of a condition. This gives women accurate information on the pregnancy which they carry. Most women are relieved when they receive a negative result. Some women are faced with a positive result, and this presents two options; termination of pregnancy, or continuation. A significant yet small number of women choose diagnosis for preparation only, with no intention to terminate in the case of a positive result (Antenatal Down Syndrome Advisory Group, 2007). This must be weighed against the risks of the procedure, the most important of which is the risk of spontaneous miscarriage.
2.1.1 The risk of spontaneous miscarriage

Amniocentesis and CVS both obtain material which is not readily accessible. The tests are therefore *invasive* diagnostic procedures (IDPs), and as with any invasive test there are extra risks involved. The most notable of these is the risk of spontaneous miscarriage. The loss of a pregnancy is a significant event, as it is the loss of a potential life. Miscarriage can have profound consequences for the whole family, with most women experiencing grief and guilt (Frost, 1996; Lee, 1996). Because of this, amniocentesis and CVS should not be used unnecessarily.

The level of risk which these IDPs pose to the fetus has been well researched. Recent literature suggests that second trimester amniocentesis increases the risk of spontaneous miscarriage by around 1% over the natural (background) risk (Alfirevic et al., 2009). As shown in Figure 7 there is significant heterogeneity between studies that have assessed miscarriage risk.

<table>
<thead>
<tr>
<th>Author</th>
<th>Spontaneous abortions</th>
<th>Difference in rates (95% confidence interval)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tabor (1980)</td>
<td>1.7% [38/2242]</td>
<td>0.0% (0.3%; 1.3%)</td>
</tr>
<tr>
<td>Andreassen (1989)</td>
<td>2.3% [38/1318]</td>
<td>0.8% (-1.3%; 1.9%)</td>
</tr>
<tr>
<td>Tongsong (1998)</td>
<td>1.8% [36/2045]</td>
<td>0.3% (-0.4%; 1.1%)</td>
</tr>
<tr>
<td>Antsaklis (2000)</td>
<td>2.1% [79/3696]</td>
<td>0.0% (0.1%; 1.2%)</td>
</tr>
<tr>
<td>Eddleman (2006)</td>
<td>1.0% [21/2096]</td>
<td>0.1% (-0.3%; 0.3%)</td>
</tr>
<tr>
<td>Kong (2006)</td>
<td>1.7% [58/3465]</td>
<td>0.0% (0.1%; 1.4%)</td>
</tr>
<tr>
<td>Olobo (2008)</td>
<td>0.97% [113/11,695]</td>
<td>0.13% (-0.07%; 0.20%)</td>
</tr>
</tbody>
</table>

*Figure 7 Risk of spontaneous miscarriage from amniocentesis. Figure shows the spontaneous abortion rate of cases (who received amniocentesis) compared with controls (who did not receive amniocentesis). The difference in rates indicates the risk of spontaneous miscarriage from amniocentesis. References are not included in reference list. Image adapted from www.uptodate.com*

CVS is less safe than second trimester amniocentesis, but only marginally (Alfirevic et al., 2009). However, it has the benefit of earlier diagnosis, from 10 weeks gestation. Transabdominal CVS is likely to be safer than transcervical CVS (Alfirevic et al., 2009). It is possible that current procedure-related losses are lower than those recorded in Figure 7, and may be due to increased use of preventative techniques, more experienced technicians and overestimation by previous studies.

Major risks apart from spontaneous miscarriage include rupture of membranes, fetal injury and infection. Complications directly to the mother are rare and have an
incidence of less than 0.1% (American College of Obstetricians and Gynecologists, 2007b).

Because of the complications, it is important that amniocentesis and CVS are only used when required, and that the mother has full knowledge of all risks posed. Non-directive counselling should be offered to discuss the test's purpose, complications, accuracy, limitations, and time required before results become available (Up To Date, 2012a). To supplement counselling, information in the form of leaflets and pamphlets should be distributed to all those given the option of the procedure.

Before proceeding with amniocentesis or CVS the risks and benefits of the procedure should be understood by the patient. This usually requires some element of counselling both before and after results are delivered. This is especially important because of the increased risk of spontaneous abortion, and because the procedure does not directly benefit the health of the mother or the fetus, instead giving information which allows options otherwise unavailable.

2.2 Prenatal screening

The complications of both amniocentesis and CVS are not common, but are potentially severe. The allocation of these procedures is therefore of utmost importance. ‘Screening’ tests aim to distribute these procedures among those most in need.

Screening tests have become popular in a number of countries to detect abnormality, and/or prevent morbidity and mortality from a disease process. Defined by World Health Organisation (WHO) criteria, “screening is the presumptive identification of unrecognised disease or defects by means of tests, examinations, or other procedures that can be applied rapidly” (Wilson & Jungner, 1968, p. 11).

Screening tests are utilised for many different conditions, including cancers (such as breast, cervical, and colon cancer), infectious disease (many sexually transmitted infections) and newborn screening (treatable congenital disease, such as phenylketonuria). In these cases, screening has the capacity to allow early intervention and halt the disease process, preventing further morbidity (and possible mortality) to the individual. When screening for communicable disease, there is the added benefit of
preventing further infections, thereby producing a direct community-wide benefit as well.

Prenatal screening for DS (and certain other conditions) differs in that treatment is unavailable for many of the conditions tested. Therefore, the function of screening is not to enable prompt treatment, reducing morbidity and mortality. The primary aim of screening tests for DS is to detect women who are at increased risk of having a fetus with a congenital disorder. This informs women about their pregnancy and provides options, such as the choice to have a diagnostic test. Screening tests are currently available in many first-world hospitals (Saller, 2008).

A screening programme organises testing on a wide scale through a recognised framework, and provides further benefits. The primary aim of screening programmes for DS is to reduce the number of unnecessary invasive diagnostic procedures, as these have an inherent risk of fetal loss (Antenatal Down Syndrome Advisory Group, 2007). Other benefits include being able to improve the information distributed to patients and practitioners, reinforce processes for informed consent, improve the safety and quality of services, and evaluate the effectiveness of the programme (Antenatal Down Syndrome Advisory Group, 2007). A nationwide programme has further benefits, allowing all women equitable access to screening. Nationwide screening programmes for DS are currently used in many countries, including New Zealand, Australia, the Denmark, and the UK.

2.2.1 Screening terminology

Screening tests are different to diagnostic tests in that they cannot confirm or rule out disease. However, they do sort patients into those at ‘increased risk’ and those at ‘low risk’. Those at increased risk are usually offered a diagnostic procedure, whereas those at low risk will not be followed up, as it is unlikely that they have the disease in question.

Screening tests aim to pick up as much as possible of the condition being tested for. Unfortunately, by detecting as much disease as possible some patients will test as increased risk, though they do not actually have the condition. These results are ‘false positives’. Other patients will test negative, though they do in fact have the condition. These results are ‘false negatives’. 
Generally, screening tests have a low false negative rate, to detect as much disease as possible. False negative results are problematic as they lead to a sense of false security, and lack of opportunity to be referred for a diagnostic procedure. False positive results are problematic, too, as they lead to increased anxiety and unnecessary diagnostic procedures.

False positive results are especially problematic when screening for prenatal conditions. This is because the invasive diagnostic procedures (IDPs) which follow a positive result have an associated miscarriage risk. If large numbers of pregnancies are labelled as being at increased risk of DS when they do not have a DS pregnancy, many women will unnecessarily choose an IDP. On a population-wide scale, this can result in significant numbers of pregnancies being lost. An effective screening programme for DS, therefore, must have a low false negative rate and a low false positive rate.

Because screening can result in such significant outcomes, policymakers have a responsibility to select the optimum programme for their population. Thankfully, many methods of screening for DS are available.

2.2.2 Methods of screening for Down syndrome

Because it has been recognised for some time that increasing maternal age is related to an increased risk of fetal aneuploidy, maternal age was the first ‘screening’ method used by maternity carers. In many countries, any pregnancy where the mother would be aged 35 or over at the expected time of delivery was treated as being at ‘increased risk’. These women would be offered amniocentesis. Those under 35 were considered at low risk of fetal aneuploidy. The age of 35 was chosen as at this age the risk of having DS pregnancy roughly equalled the then-accepted risk of spontaneous miscarriage from amniocentesis.

This method of ‘opportunistic’ or ‘ad hoc’ screening has since been abandoned in New Zealand, the United Kingdom (UK), and many other countries. It is now recognised by professional bodies such as the American Congress of Obstetricians and Gynecologists (ACOG), and the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG), that all women should be offered prenatal screening regardless of age (American College of Obstetricians and Gynecologists, 2007a; The Royal Australian and New Zealand College of Obstetricians and
Gynaecologists, 2007). There are several reasons for this. Firstly, women of all ages are susceptible to fetal abnormality. Secondly, many women have differing perceptions of risk, and this is especially pertinent in reproductive decisions. Lastly, without screening most neonates with fetal anomalies are born to women under the age of 35, simply as the birth rate in this cohort is higher (Collins, 2008).

Thus, screening methods other than maternal age alone must be used. Though there are many methods available, most measure levels of certain markers in the pregnant woman’s blood. One exception is Nuchal Translucency (NT) scanning, which uses ultrasound to measure the thickness of the developing fetus’ neck to predict the likelihood of DS. These techniques are non-invasive and do not have significant complications in the same way that amniocentesis and CVS do. But because they are non-invasive and do not source material from the fetus itself, they are not accurate enough to be considered diagnostic. Widely used screening methods for DS are shown in Figure 8.

<table>
<thead>
<tr>
<th>Nuchal translucency (NT) measurement</th>
<th>The width of the translucent space at the back of the fetal neck determined by ultrasound.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Combined test</strong></td>
<td>First trimester test based on sonographic and maternal serum measurements: NT, β-human chorionic gonadotropin (β-hCG: free or total), and pregnancy-associated plasma protein-A (PAPP-A), together with maternal age.</td>
</tr>
<tr>
<td><strong>Triple test</strong></td>
<td>Second trimester test based on measurement of maternal serum alpha-fetoprotein (AFP), unconjugated estriol (uE₃), and β-hCG (free or total), together with maternal age.</td>
</tr>
<tr>
<td><strong>Quadruple test</strong></td>
<td>Second trimester test based on measurement of maternal serum AFP, uE₃, β-hCG, and inhibin-A, together with maternal age.</td>
</tr>
<tr>
<td><strong>Integrated test (full)</strong></td>
<td>The integration of measurements performed during the first and second trimesters into a single test result. Typically, the integrated test refers to the integration of NT and PAPP-A measurements in the first trimester with the quadruple test markers in the second trimester, together with maternal age.</td>
</tr>
<tr>
<td><strong>Serum integrated test</strong></td>
<td>A variation of the integrated test using maternal serum markers only: PAPP-A in the first trimester and the quadruple markers in the second trimester, together with maternal age.</td>
</tr>
</tbody>
</table>

Figure 8 Commonly used methods for Down syndrome screening. Image adapted from www.uptodate.com
In most models, empirical results are combined with information on the mother such as smoking status, gestational age, and maternal age. Sometimes, a number of serum markers from the woman’s blood are combined to give a more accurate result. Of particular note is the combined test, which uses both serum markers and NT results to give a combined risk estimate of DS in the fetus.

Different screening tests are better suited for different stages of pregnancy. Some, like the combined test, are better suited for testing within the first trimester. Others, such as the triple and quadruple tests, are more effective in the second trimester. The integrated and serum integrated tests require two tests, in both the first and second trimesters. As women choosing screening prefer to have as early testing as possible (Mulvey & Wallace, 2000), testing in the first trimester can be preferable to testing in the second trimester. It can enable more time for decision-making, with the option of an earlier abortion if chosen.

All of the tests shown in Figure 8 can detect other congenital disorders, such as trisomy 13, trisomy 18, spina bifida, and many rare metabolic disorders (National Screening Unit, 2009; Saller, 2008). Screening which employs ultrasound (such as the NT measurement) may detect major structural anomalies in the fetus, such as skeletal anomalies and abnormalities of the heart, renal tract and gastrointestinal system (National Screening Unit, 2009). Thus while DS is the most prevalent condition screened for, tests for DS typically detect other conditions as well.

As the tests measure different variables they vary in their accuracy. There have been several studies comparing the effectiveness of these screening methods, the two largest of which are the First and Second Trimester Evaluation of Risk Trial (FASTER) and Serum, Urine and Ultrasound Screening Study (SURUSS) (Stone & Austin, 2006). Results from these trials are shown in Figure 9.
Figure 9 Results of the FASTER and SURUSS studies. Detection rate = the percentage of fetuses with DS detected as being at ‘increased risk’. FPR = false positive rate. ‘Procedure related losses’ are the spontaneous miscarriages from women who go on to receive amniocentesis or CVS after receiving an increased risk result. Image adapted from www.uptodate.com

The FASTER and SURUSS results indicate that the integrated and serum integrated methods of DS screening are the most accurate, because they have the lowest false positive rate at a given detection rate. Integrated screening is currently used in the US and parts of Europe. Integrated methods are problematic for a nationwide screening programme, as they require two separate tests, with results and the option of diagnostic testing withheld until the second trimester (McEwan et al., 2012). This is especially problematic for women at high risk of a DS pregnancy. Therefore combined testing is the favoured method for population-wide screening in many countries, such as in the UK and New Zealand (McEwan et al., 2012).

Since the FASTER and SURUSS trials were conducted a new method of screening has been developed named contingent screening (Cuckle et al., 2008). This is based on integrated screening, but separates results into three categories; those at ‘high risk’, ‘low risk’, and ‘intermediate risk’. Those at high risk are offered invasive diagnosis immediately, while those at low risk leave the screening pathway. Those at intermediate risk are offered a second trimester test, as in the integrated method. Contingent screening has been found to have the best cost effectiveness compared to other screening methods (Gekas et al., 2009). Despite this, policymakers are unlikely to change screening programmes in the near future due to the complexity they introduce, which may prove confusing for both maternity carers and pregnant women (McEwan et al., 2012).
2.2.3 The screening pathway

The DS screening pathway for all pregnant women in New Zealand is shown in Figure 10. The pathway sorts women into two categories, dependent on gestational age and the availability of ultrasound. Women less than 14 weeks pregnant who can receive ultrasound are offered first trimester combined screening. Those who are unable to access ultrasound, or are more than 14 weeks pregnant, are offered the quadruple test, which does not involve a NT scan and only uses a blood test. Women do not receive screening more than once in pregnancy (i.e., combined screening would not be supplemented with quadruple screening after 14 weeks).
The pathway shown in Figure 10 is very similar to the process used in the UK, which also uses combined and quadruple screening measures (McEwan et al., 2012). In the UK, the threshold for an increased risk result is a one in 150 chance that the fetus has DS (McEwan et al., 2012). In the case of an increased risk result, women are provided with a pamphlet explaining their options, together with the offer of referral to an obstetrician to discuss diagnostic testing.
Regulated screening programmes like those used in New Zealand and the UK allow more rigorous standards to be put in place for a testing process. These standards affect all women, not only those at certain hospitals. As prenatal screening is sufficiently different to other forms of disease screening, it is especially important that procedures surrounding informed consent are robust. For this to occur, women faced with screening must have a clear understanding of the decisions which may need to be made at future steps of the screening pathway. Information must be accurate and relevant, the right to decline screening emphasised, and any decision respected. Women should have time to ask questions, talk to others and reflect on choices at each step in the pathway. These guidelines are echoed in those which currently inform prenatal screening in New Zealand (National Screening Unit, 2009).

Similar guidelines inform screening for DS in the UK. Written and verbal information regarding DS screening must be received by the woman at least 24 hours before making a decision (McEwan et al., 2012). Reviews of those carrying out screening (such as sonographers performing NT scans) as well as data gathered on the processes surrounding testing are regularly synthesized and reviewed to ensure best practice.

2.2.4 Evaluating screening

When evaluating a screening process, the New Zealand Ministry of Health suggests that the following criteria are followed. These are in line with criteria suggested by, among others, the United Kingdom National Screening Committee.

Criteria for assessing screening programmes:

1. The condition is a suitable candidate for screening.
2. There is a suitable test.
3. There is an effective and accessible treatment or intervention for the condition identified through early detection.
4. There is high quality evidence, ideally from randomised controlled trials, that a screening programme is effective in reducing mortality and morbidity.
5. The potential benefit from the screening programme should outweigh the potential physical and psychological harm (caused by the test, diagnostic procedures and treatment).
6. The health care system will be capable of supporting all necessary elements of
the screening pathway, including diagnosis, follow-up and programme
evaluation.

7. There is consideration of social and ethical issues.

8. There is consideration of cost-benefit issues. (National Health Committee,
2003, p. 3)

Though helpful for diseases such as cervical cancer and newborn screening,
some of the criteria are not suitable for all types of screening. Specifically, the third and
fourth criteria are inappropriate for DS screening, as noted by a report commissioned by
the National Screening Unit of New Zealand (Antenatal Down Syndrome Advisory
Group, 2007). Prenatal screening is not offered so that DS can be treated, but for women
“to make informed decisions about their pregnancy, with appropriate support and
counselling” (Antenatal Down Syndrome Advisory Group, 2007, p. 50). These
interventions are not necessarily in the best interests of the woman, and it is up to her to
decide. Options available include whether to have a diagnostic test, whether to continue
or terminate the pregnancy, and preparing for the birth and life of a child with DS

Criterion four is also unsuitable for screening for DS. If the purpose of screening
is to reduce the live-birth prevalence of DS, this will inevitably restrict a woman’s
independent choice regarding screening and diagnosis. Women would be encouraged to
test, and (possibly) abort, as this would reduce the births of newborns with DS.
Moreover, such an approach may send the message to people with DS that they are
undesirable and a ‘drain on society’ (the validity of this claim will be dealt with in
Chapter 4). For these reasons the purpose of screening should not be to reduce the
morbidity and mortality of DS, but to reduce the harm from unnecessary diagnostic
procedures and allow decision-making for all (Antenatal Down Syndrome Advisory
Group, 2007).

Criterion eight suggests that cost effectiveness should be taken into account
when assessing screening programmes. Based the most recent estimate which used
evidence gathered in 1988, the total lifetime economic costs of a person with DS in the
US was estimated to be around $410,000 US Dollars, not adjusted for inflation
(Waitzman et al., 1994). This encompasses support care, special education, and
healthcare for those with DS over that of the standard population. This cost was calculated for California, and costs will be different in other countries.

But is cost effectiveness a valid criterion to assess prenatal screening? With screening for cancer or infectious disease, diagnosis and treatment are often regarded as responsible follow-up procedures. For DS there is no such judgement; though screening allows invasive testing it does not mandate it, nor does it mandate the abortion in the case of a positive result. Assessing prenatal screening in terms of the ‘savings’ from lives with DS ‘prevented’ over the total cost of the programme can be misleading, as preventing people with DS should not be the aim of the programme.

However, though cost-benefit analysis can be misleading does not make it wrong. The criteria shown above ask us to give consideration to cost-benefit issues, but this is not the only consideration. Indeed, cost-benefit analysis tells us nothing as to whether or not a practice is justified (for this is the aim of this thesis). But when implementing any policy, the actual costs of the programme (tests, follow up, diagnosis) are important to consider. Though cost-effectiveness is a valid consideration its helpfulness is limited; and it is by no means our only concern.

Apart from criteria three and four, all other criteria developed by the National Health Committee (2003) are valid considerations to assess a programme which screens for DS. In addition to those noted above, separate criteria have been suggested specifically for prenatal screening. Suggested requirements include that participation must be strictly voluntary, that informed consent must be given and based on sound and easily-understood information, and that several options are open to help women make or act on decisions (Antenatal Down Syndrome Advisory Group, 2007). These criteria are helpful, and form appropriate concerns to assess both current and future screening programmes.

2.3 Outcomes of prenatal testing

2.3.1 Individual outcomes

Though the aim of prenatal testing (both screening and diagnosis) is not necessarily to terminate a DS pregnancy, this is often the outcome of such tests. This is
because the majority of women abort when they receive a positive result from amniocentesis or CVS.

Estimates of the termination rate vary, and probably depend on religious, cultural and demographic factors of the study population. A systematic review by Jaime Natoli (2012) included 24 studies published between 1995-2011 which were conducted in the US. Her findings indicated that the mean rate for termination following a positive diagnosis of DS was 67%. There was significant heterogeneity between studies, probably reflecting varying societal factors in differing study populations. Several studies included in the review suggested that termination rates had decreased in recent years.

That termination rates may be reducing is corroborated by an earlier review which used data collated from studies between 1980 and 1992 (Mansfield, 1999). Caroline Mansfield (1999) concluded that 92% of pregnancies which received a positive diagnosis for DS were subsequently terminated. This study had an international focus, sourcing data from studies conducted in the UK, New Zealand, Australia, US and continental Europe.

Termination rates likely reflect various socioeconomic, religious and cultural factors, as the individual decision to abort a fetus with DS encompasses many values (Bryant et al., 2006; Garcia et al., 2009). That termination rates may be reducing could be a reflection of the increased life prospects for those with DS, and better financial, social, and educational support for their families (Davidson, 2008; Feingold & Geggel, 2001). Moreover, it could be a factor of a reduction in stigma against DS, less medically orientated information given to those with a positive diagnosis, or point to the success of non-directive counselling emphasising the role of individual choice. Greater numbers of women may be choosing prenatal diagnosis to prepare emotionally for the birth of a child with DS, or make specific delivery plans, instead of planning on termination in case of a positive result (Natoli, 2012).

However, termination rates remain high. Several explanations for this are given. Most significantly, those who elect to have a diagnostic procedure are a self-selecting population (Natoli, 2012). Women who choose amniocentesis differ from women who decline, in their attitudes about pregnancy termination, perception of amniocentesis-associated miscarriage risk and their need for certainty about whether the fetus has DS.
(Kobelka et al., 2009). As the risk of miscarriage is a significant consideration, it dissuades many women from using invasive diagnosis only for preparation. Therefore it is reasonable to assume that many women choose diagnosis with the intention of terminating in case of a positive result. Thus the number terminating when a positive result is received would be expected to be high, and is not necessarily a reflection of health professionals encouraging an end to pregnancy.

2.3.2 Reasons for termination

Why, then, do women abort in the case of DS? Only one study has examined women’s reasons for pregnancy termination. Korenromp et al. (2007) examined the decisional processes of 71 women who had terminated a DS pregnancy four months earlier. Eighty-three per cent of women terminated because they believed that the ‘burden’ [sic] of DS was too heavy for the child, and 73% believed it too heavy for other children. Extensive findings from the study are found in Figure 11.

![Figure 11: Motivations for termination of a Down syndrome fetus. Image adapted from Korenromp et al. (2007, p. 149)](image)

Korenromp et al’s (2007) findings indicate that both self-interested and non-self-interested motives play a role when considering the termination of a fetus with DS.
Twenty-one per cent of respondents expressed high levels of doubt about the decision. A woman’s partner was the most likely to be perceived as influential in the decision process regarding termination, being important in 74% of cases. Obstetricians, family and friends, and geneticists were also likely to be prominent in the decision.

Ninety-seven per cent of women were happy with their decision, indicating that they would make the same decision again. Some pressure to terminate was felt by 14% of women, similar to the 17% found by an earlier study (Korenromp et al., 2006). This pressure was likely to be from the ‘moral values of society’, family and friends, or medical staff. Though the number of women receiving pressure to terminate by medical staff was low (7%) it is problematic, as it is not consistent with non-directive counselling.

2.3.3 Collective outcomes

Though the aim of prenatal testing is not necessarily to decrease the number of births with DS, this is invariably what occurs when such tests are available and a significant proportion of women desire to abort. Worldwide however, the proportion of DS births has not decreased (Cocchi et al., 2010). Cocchi et al. (2010) analysed birth data from 1993-2004 from 14 developed nations. While there was a marked decrease in DS births in France and Italy, there was an increase in Canada, Italy, Israel and Norway. Taking all countries into account, there was no significant change in the overall proportion of births with DS. Similar patterns of no significant change were found in Victoria, Australia (Collins, 2008), and an increase in live birth prevalence in the US (Besser et al., 2007).

Although the proportion of DS births has not decreased it is clear that the number of terminations of fetuses with DS has risen worldwide (Cocchi et al., 2010). This is due to the increased availability of prenatal testing (Cocchi et al., 2010). That there has not been a drop in the prevalence of DS at birth is due to a third factor: there has been a worldwide increase in the conception of fetuses with DS. As discussed in Chapter 1.2, advanced maternal age is a significant risk factor for conceiving a fetus with DS. Hence, while the use of testing has increased worldwide, so has maternal age (Cocchi et al., 2010), leading to the surprising finding that there not been a significant change in the live birth prevalence of DS.
Worldwide, the ways in which prenatal testing is offered to women differs. In some countries screening is only offered to older women, whereas in others screening is offered to all. Many of the countries used by Cocchi et al. (2010) did not have a nationally recognised screening programme. It is therefore hard to estimate the impact of a widespread screening programme such as those now used in New Zealand, the UK, and Australia using the Cocchi et al. data.

Ekelund et al. (2008) assessed the impacts of the introduction of a screening programme in Denmark between 2004-2006, using data on live births from 2000-2007. From 2004 all pregnant women were offered the combined DS screening test in the first trimester of pregnancy. The number of infants born with DS nationwide ranged from 55-65 per year in the five years previous to the introduction of the screening, but only 31 and 32 in the two years following. Thus the introduction of screening had the effect of reducing DS births by around 50%, though this was not specifically the aim of the programme. Similar effects could be expected from screening programmes in Australia and New Zealand, though the impacts of these have yet to be comprehensively evaluated.

In many countries therefore, the rise in prenatal testing has not resulted in reduced birth prevalence of DS. It is difficult to foresee whether the worldwide birth prevalence of DS will drop. This is because there has been a steady rise in maternal age observed in many countries, which is likely to continue for the near future (Royal College of Obstetricians and Gynaecologists, 2009).

It is hard to predict if the worldwide rise in maternal age will level off. In New Zealand, the mean and median age of women at childbirth has remained steady at around thirty years, with a small decrease in recent years (Statistics New Zealand, 2011). Though the average maternal age is a product of many socio-demographic factors, it seems reasonable to assume that unless the accessibility, viability and efficiency of assisted reproductive procedures dramatically increases worldwide maternal age in the developed world will not continue to rise indefinitely.

Thus in some countries at least, the increase in use of prenatal testing (and therefore termination) could be expected to result in reduced births of infants with DS, as seen in Denmark. Depending on the magnitude of the reduction, this may or may not result in fewer people with DS. Individuals with DS now live longer than ever before, and
life expectancy is predicted to increase (Bittles & Glasson, 2004). At least temporarily, this would reduce the magnitude of any change in the proportion of those with DS in society.

In light of these considerations, a significant decrease in the numbers of those with DS requires a marked increase in the uptake of testing. With this in mind, what new technologies may markedly increase uptake rates?

### 2.4 Future prenatal testing

Current testing for DS involves either non-invasive screening techniques, such as blood tests or ultrasound, or invasive diagnostic tests, such as CVS or amniocentesis. Future methods of testing would be both non-invasive and diagnostic; they would be an example of non-invasive prenatal diagnosis (NIPD).

In 1997 it was discovered that 3-6% of cell-free maternal DNA is of fetal origin (Lo et al., 1997). These fetal DNA fragments can be analysed to detect specific conditions. NIPD for DS is currently in development and shows promise. Indeed, an early form of NIPD for DS has been available in the US since October 2011 (Laino, 2011), though it currently lacks the precision to be considered truly diagnostic (Benn et al., 2012).

NIPD has the potential to be used for a large number of diseases and characteristics, as it samples the fetal genetic material directly. The most straightforward use of NIPD is for fetal sex determination, for it only requires detection of the presence of a Y chromosome. This can be done with very high accuracy by the tenth week post-menstruation (Scheffer et al., 2010). Testing for fetal aneuploidy is more difficult, instead needing to identify changes in chromosome proportion. Specific challenges include blocking out the normal background ‘static’ of maternally-derived maternal DNA, and the relatively small contribution to cell-free DNA by the fetus.

To test for DS, NIPD analyses the proportions of fragments of the 21st chromosome compared to other chromosomes in maternal plasma to determine whether the fetus has trisomy 21 (Laino, 2011). As the fetus has three copies of the chromosome instead of the usual two, a greater number of fragments would be expected in maternal blood. At present, the most reliable method to measure these proportions is
massively-parallel shotgun sequencing, which sequences short segments of cell-free DNA. It then generates the distribution of this DNA to confirm or rule out DS (Chiu et al., 2008). Other methods, such as chromosome-selective sequencing (Ashoor et al., 2012), DNA methylation comparisons (Chim et al., 2008), and mRNA sequencing (Lo et al., 2007) also show promise.

The primary benefit of NIPD would be the avoidance of fetal loss associated with amniocentesis and CVS. Though the increased risk of loss from these procedures is low, when these are offered on a large scale the number of spontaneous miscarriages may be significant. It has been estimated that, even when DS screening is in place, around 400 pregnancies in England and Wales are lost every year due to invasive diagnosis (Buckley & Buckley, 2008). Using NIPD these losses would not occur.

NIPD also offers other benefits. At present it can be used from 10 weeks of pregnancy (Palomaki et al., 2012), although it holds the potential to be used from seven weeks. This is because fetal nucleic material is present in maternal blood very early in gestation (Schmitz, 2012). Although CVS can be used from 10 weeks, amniocentesis is more readily available, possibly safer and should be used after 14 weeks gestation (Royal College of Obstetricians and Gynaecologists, 2010). Results from current diagnostic procedures take from one to three weeks to become available (National Screening Unit, 2009), and results are often received after 17 weeks of pregnancy (Skotko, 2009). NIPD, therefore, would enable more time for decision-making, enabling an earlier and safer termination of pregnancy if desired. Indeed, NIPD could allow more parents to test to prepare for the birth of a child with DS, as unlike invasive diagnosis there is no potential for major complications.

There is evidence that NIPD is a test which is attractive to patients. A study in the US by Tischler et al. (2011) found that 72% of pregnant women express an interest in receiving the test. Safety of the fetus was the most important factor influencing their decision. Because of this, the timing of the results and the relative ease of the procedure, NIPD could be the future test of choice for prenatal testing, supplementing or even replacing screening and invasive procedures within the next 10 years (Greely, 2011).

Current forms of NIPD should only be considered as very accurate screening tests (Benn et al., 2012). Future forms of NIPD may always be limited in the detection of some disorders, such as spina bifida. However, NIPD could be offered in tandem with current
screening methods, and need not replace them. Indeed, NIPD was recently shown to be effective in detecting trisomy 13 and trisomy 18 (Palomaki et al., 2012).

With time, costs of NIPD should reduce and accuracy may improve, such that it may be offered on a wide scale (Chitty et al., 2012). Thus, it is not unreasonable to predict that in the future, many women may use NIPD and more women may terminate DS pregnancies than at present. Though life expectancy of DS and the age of mothers may increase, it is possible that the number of those with DS will decrease in the future. The ethical implications of this are explored in Chapter 5.3.

### 2.5 Conclusion

Prenatal testing for DS has been available for women of advanced maternal age since the 1960s, and for all pregnant women in recent years. Though invasive diagnostic techniques were the only method of detection at first, advances in ultrasound and the measurement of serum markers means that many screening methods are now available. Nationwide screening programmes currently operate in several developed countries, and screening tests themselves are offered in many more. It is likely that with its avoidance of miscarriage risk, NIPD may become the future prenatal test of choice. Whether or not this will make DS a rare disorder is uncertain, but looks unlikely for the near future.

Although prenatal testing is used worldwide, this does not mean that its use is justified. The primary aim of diagnosis is to enable an option otherwise unavailable; termination, in the case of a positive result. But is a termination of pregnancy in this way permissible? Or are women acting unreasonably, selfishly, and immorally? It is to this question that we now turn.
Part Two: A discussion of ethics
Chapter 3 AN INDIVIDUAL CHOICE: THE MORALITY OF ABORTION FOR DOWN SYNDROME

The aim of prenatal testing is to confirm the presence or absence of DS in the fetus of a pregnant woman. A diagnosis of DS allows the option of elective abortion. Though there are other benefits arising from testing, such as psychological preparation for the birth of a child with DS, and reassurance in the case of a negative result, these outcomes are secondary. Thus, when evaluating the moral permissibility of prenatal testing for DS the first question we must ask is this: is the abortion of a fetus with DS morally permissible? And how does the abortion of a fetus with DS compare to an abortion for ‘any’ reason?

3.1 The morality of abortion

Abortion has been practised in many cultures throughout history (Devereux, 1967), but its past use was limited by both danger and social stigma (Joffe, 2009). Now, nearly three-quarters of the world’s women live in countries where abortion is legal; either in all circumstances, or when specific medications or conditions are present (Boland & Katzive, 2008).

The morality of abortion is emotive and far from resolved, although the debate has existed for nearly a century. The primary point of contention is the moral standing of the fetus; this is sometimes described as the fetus’ ‘right to life’ (Gillon, 2001). Those who discuss the morality of abortion ask if the fetus has a right to life, at what stage it gains this right, and if this right outweighs the mother’s wish to end a pregnancy. There are a number of positions which may be held on the permissibility of abortion; among others, these include those who assert that it should be allowable in all cases, those who believe it should be subject to certain restrictions, and those who oppose its use altogether.

I have chosen to align myself with a gradualist approach to abortion, “which suggests that the developing fetus should be regarded with increasing moral status as pregnancy progresses” (Shakespeare, 2006, p. 94). This position permits abortion at earlier stages of pregnancy, but requires increasingly strong reasons as pregnancy progresses. This position is useful as it mirrors that which informs the legislation found
in many jurisdictions, and thus the restrictions which govern many women. It recognises that a woman’s right to choose whether to be a parent is limited by the increasing right to life of the developing fetus. This position seems reasonable, as it recognises that reproductive autonomy is limited in regards to other considerations.

Under the gradualist approach, abortion is morally permissible until later in pregnancy. The limit where abortion is thereafter impermissible is recognised to be around 20-24 weeks in many countries. The following discussion is based on this premise.

3.1.1 Reasons for abortion

Abortions may be carried out for any number of reasons. These reasons tend to fit into two categories; they are either ‘medical’ reasons, or ‘social’ reasons. Medical reasons include if the continuation of pregnancy will result in life-threatening harm to the mother (such as in renal failure, or severe cardiac disease), or the presence of a significant pathology in the fetus (such as spina bifida, or DS). The latter can be labelled a ‘selective’ abortion, as a select, specific trait in the fetus is viewed as sufficient for termination of pregnancy. Social reasons encompass abstract qualities of the social positioning of the mother: such as living in poverty, or already having a large family, or carrying an unplanned pregnancy.

The most cited reasons for abortion are a lack of marital or family support, the cost of raising a child, and that a baby will halt the fulfilment of responsibilities to work, education, or dependents (Finer et al., 2005). These are not traits but circumstances of the mother. An abortion on these grounds is a ‘social abortion’. It is of note that either social or medical reasons must be proved as constituting a harm in New Zealand (Crimes Act 1961) and some other jurisdictions.

What follows is a comparison of the moral permissibility of social and selective abortions; specifically, selective abortions where the trait in question is DS. I will examine whether a fetus with DS has a greater, lesser or similar right to life when compared with a fetus without DS at the same stage of development. The criteria used will be viability, sentience, and the potential to live a good life. Each criterion will be recognised within the gradualist paradigm of increasing right to life.
3.1.2 Argument for viability

The argument for ‘viability’ regards the capacity of the fetus to survive outside the womb with adequate healthcare as a strong factor in whether or not an abortion is permissible. A fetus is defined as viable when it is able to survive independently of the pregnant woman, with appropriate intensive care. Currently, the earliest this can occur is 22 weeks of age (Gillon, 2001). Before this time, the fetus will not survive a premature birth; it is contingent on continuing pregnancy for survival.

Through this logic, a fetus’ right to life increases dramatically after this 22 week (or thereabouts) threshold. The viability criterion strongly influences legislation, shown by a restriction on abortion after 20 to 24 weeks of pregnancy in many jurisdictions. The argument for viability often influences the views of medical professionals, especially those in the field of obstetrics (Gillon, 2001).

The argument for viability is criticised as a fetus’ survivability is contingent on the care and technology available to it. Thus, while a fetus may be viable from 22 weeks in some Western intensive-care units, other hospitals cannot guarantee viability until later; and hospitals in developing countries much later still. Some argue that viability tells us little of the moral status of the fetus, as viability is determined by the presence or absence of technology (Gillon, 2001). Regardless, the viability criterion is a very practical way to approach abortion, providing a clear line where the mother’s right to terminate a dependency ends, and abortion is no longer permitted.

There is some evidence that a fetus with DS is less viable than a non-DS fetus, as they have a significantly increased miscarriage rate throughout pregnancy. Between 12 weeks and term 43% of fetuses with DS are lost or result in stillbirth (Morris et al., 1999). Newborns with DS have slightly poorer outcomes compared with non-DS newborns; 90% of neonates with DS survive to five years of age (Halliday et al., 2009). Furthermore, there are many co-morbidities associated with DS which may make the fetus unable to survive a premature birth.

Though no conclusive information is available, it seems reasonable to suggest that some fetuses with DS become viable at a later stage of pregnancy than some without DS. But whether this applies to the entire group of fetuses with DS is unclear. Furthermore, any ‘viability delay’ is probably in the order of a couple of weeks, and not
of major importance. This indicates that there is little significant difference between DS and non-DS fetal right to life on the basis of viability.

3.1.3 Argument for sentience

If a creature possesses ‘sentience’ then it has the capacity to have experiences, including that of pain (Warren, 1993). For many of us, our regard for sentience informs our treatment of other animals and plants, regarding the destruction of a patch of grass as less wrong than the killing of a fly, and how these both differ to the killing of an ape. Under this approach more harm is done to a being if it qualifies as being sentient.

It is plausible that for a creature the capacity for sentience is related to neural capacity. Neural capacity is determined by functions such as brain size, the complexity of neural networks, and most importantly for the fetus, neural development. The threshold for sentience will vary, but it is likely that around the end of the second trimester the brain is sufficiently developed for the fetus to be regarded as sentient (Gillon, 2001).

Neonates with DS show marked reductions in the volume of several brain regions, and there is evidence that aberrant neural development occurs at the fetal stage (Dierssen, 2012). Children with DS show considerable learning delay, and it seems reasonable to assume that a fetus with DS has a neural ‘delay in sentience’ compared with the non-DS fetus. However, in a similar way to the viability criterion, this delay is probably not hugely significant. As our understanding of what constitutes sentience in the fetus is uncertain (Gillon, 2001) and our knowledge of the fetal brain with DS limited (Dierssen, 2012), any sentience disparity does not seem a strong criterion with which to value a fetus without DS more highly than a fetus with DS.

3.1.4 Argument for potential

Some propose that it is the potential of the developing fetus to become something of great value- a human person with unequivocal right to life- which confers a prima facie value (Gillon, 2001). Ending this process prevents the life of a human with full moral status, and therefore all abortion is wrong.

The argument for potential is criticized on its logical implausibility; for as John Harris has pointed out “we all have the potential to become corpses” (cited in Gillon, 2001, p. 6). But Harris’ objection is partially flawed, for nearly all cultures respect and
honour the bodies of the dead, whilst not treating them as actual persons. Human embryos are treated as valuable and deserving of respect though they may be used for research, or ultimately discarded (Bridge, 2012). Indeed, human material is often treated carefully though it may not have the direct potential to develop into human beings—such as donor organs, or harvested eggs for in vitro fertilisation (IVF).

Though not incontrovertible, the potential of the fetus is undeniably important when assessing fetal right to life. How, then, does the potential of a fetus with DS compare with one without DS? In the following discussion I do not confine myself to the conventional definition of potential as the potential to become a human being, as both DS and non-DS fetuses will achieve this. Instead, I re-cast the argument for potential in a new light, assessing each fetus by ‘the potential to live a good life’.

3.1.5 Down syndrome and ‘the good life’

Framed in this way, the question of potential is problematic to answer, as it requires value to be attributed to what is most necessary for a ‘good life’. These judgements strike many as extremely subjective, and some may feel that it is inappropriate and wrong to attribute value to concepts which only a few may hold in high regard. Nevertheless, several scholars have attempted to answer this question.

Jonathon Glover (2006) proposes several models encompassing attributes required for a good life. He regards the good life as one where the capacity for human flourishing is not impaired. Glover’s idea of human flourishing is derived from the Greek term ‘eudaimonia’, used predominantly in virtue ethics and standardly translated as ‘happiness’ or ‘wellbeing’ (Hursthouse, 2012). Glover does not regard this as an ‘all or nothing’ phenomenon; that is, there is no threshold for a good life because some lives are more ‘good’ than others.

Glover (2006) offers us several ways to assess a life. A ‘Darwinian’ view, he proposes, would take into account only the functions required for survival and reproduction. A ‘normal-functioning’ view would require an assessment of the standard functioning of physical and psychological systems. Both of these approaches are too restrictive, and reject the fact that many who live with a loss of functioning lead good lives. Though objective, these accounts give no weight to the subjective experience of persons, which are surely important when considering if one leads a good life.
A further model with which to evaluate the good life is espoused by Martha Nussbaum (1998). Her approach is pluralistic, citing many qualities - health, shelter, nourishment and sex, together with social interaction, love and self-reflection, among others - as integral for the good life (Glover, 2006). This approach can be thought of as a form of liberal Aristotelianism, for it was Aristotle who first championed the notion of the balanced life through his concept of the ‘Golden Mean’ (Budd et al., 2004). Nussbaum’s approach to the good life is helpful as it encompasses both subjective experience and the presence of objective states. But because this list is so long, it gives no indication of the relative importance or desirability of individual qualities or states of mind. For example, the Aristotelian philosophy tells us nothing of the desirability of love when compared to self-reflection, or if health or sex is more necessary for the good life. This philosophy indicates that all these qualities contribute to the good life, but does not tell us which are more important than others.

Lastly, Glover (2006) presents a utilitarian approach, assessing a life on how desirable it is by the presence of happiness and the absence of pain. But this method is also limited in its usefulness, shown when this is taken to its extreme; for a life of pure pleasure does not suggest a good life at all, but a life devoid of meaning.

Glover (2006) therefore proposes that we should pursue a ‘two-fold’ path when predicting or assessing one’s life. This method requires an examination of both subjective experience (how the subject feels about their life, e.g. frustrated or content) and objective measures (which could include the presence of the Aristotelian ‘goods’ mentioned previously). These two measures are related, but not identical, as the case of disability shows.

Glover (2006) defines disability as a “functional limitation which impairs the capacity for human flourishing” (p. 9). Though he concedes that those with disability may subjectively lead good lives, he asserts that due to the objective, irremovable limitations set by impairment these lives are less good than others when assessed by his ‘two-fold’ approach. That is, people with DS live lives which are enjoyable and happy, and we respect and value their rights as much as those of any other individual, yet because DS precludes significant options it is undesirable:

To think that a particular disability makes someone’s life less good is not [discrimination]. It does not mean that the person who has it is of any less
value, or is less deserving of respect than anyone else. (Glover, 2006, p. 35)

Through Glover’s (2006) framework, we come to the conclusion that a fetus with DS has less potential to lead a good life than the non-DS fetus. This does not mean that all individuals with DS and other disabilities lead awful lives; as shown in Chapter 1.4, no statement could be further from the truth. Neither does Glover’s conclusion mean that all persons without disability lead good lives; due to the absence of certain ‘human goods’, social attitudes, and a disparity in subjective experience this is also not the case. People with DS can still live good lives. But for Glover, having DS is a barrier to leading the ‘most good’ life.

This logic is sound, if the limitations of DS are objectively undesirable. This claim is tenuous, as some may regard these limitations as fundamentally unimportant. Objections can be made to Glover’s argument on two grounds. Firstly, one could question his conception of disability, proposing that ‘human flourishing’ is not impaired by a loss of function. Secondly, one could regard health problems, cognitive impairment and a lack of independence as not precluding ‘the good life’. It is possible to argue that Glover’s (2006) concept of objective ‘Aristotelian goods’ is misdirected, and the qualities in life that matter are such things as meaningful interaction and the ability to feel emotion, for example.

This position is equally valid. With no true, undisputed concept of the qualities required for the good life it is difficult to reach a definitive conclusion on whether a fetus with DS is more likely to experience a ‘less good’ life than a fetus without DS. Though we can agree on what constitutes a terrible life - namely, unbearable amounts of pain and suffering - people with DS experience no more of these than many other individuals.

If one believes that the impairment of DS impairs the capacity for human flourishing, then one will assert that a fetus with DS has less potential to achieve a good life. On the other hand, if one believes that these limitations are unimportant and merely challenges to be overcome like any other in life, then the fetus with DS and the non-DS fetus are equal. For the present I will regard each of these positions as equally tenable.
3.1.6 The right to an open future

A concept introduced by Buchanan et al. (2000) is the future child's "right to an open future" (p. 170), similar to that first espoused by Joel Feinberg (1980). To ensure an open future suggests a way in which parents would be morally permitted to act, since it allows the child to follow whatever path they desire, and develop as they choose.

A fetus with DS is limited in some of its future options; it has a 'less open' future when compared to a non-DS fetus. This is not to say that the life of an individual with DS is uniform and devoid of choice; that is simply not true. However, the impairment of DS does limit some options; in some cases, limiting an open future. From this logic, the potential of the fetus with DS compared to the non-DS fetus for an open future is reduced.

Support for this view is echoed in our actions which oppose the limitations of any disabling condition, in the same way that we oppose the limitations set by any unfair circumstance. For example, we aim to reduce the burden of poverty by offering social welfare, to reduce the limitations set by financial inequality. We offer scholarships for education, allowing those who would not otherwise have the option of attending institutions such as university. In the same way we aim to treat and cure, opposing the limitations disease sets on patients’ lives. Indeed, some have claimed that reproductive testing offers a way of correcting the unfairness of the natural genetic lottery, thereby ensuring genetic equality for all (Buchanan et al., 2000).

A fetus with DS could be prevented from having an open future, if one regards the impairment of DS as significant. But it should be noted that all of us are limited in our futures; not only by our genetic makeup, but also the interplay of our social and physical environment. Most parents do not act to give their children an open future, instead directing them to pursuits and activities which they believe are beneficial for the child (and sometimes, themselves). Indeed, it could be claimed that a ‘right’ to an open future entails a right which does not actually exist; for if it was to exist, it would oppose the necessary authority and direction which constitutes good parenting.

Though a child’s right to an open future can be disputed, it seems to constitute a valid reason as to why a parent’s decision for selective abortion would be morally permissible, or more permissible than that of a social abortion. Though it is clear that a
fetus with DS is not harmed in being born and living, it is probable that most women see the choice of termination as choosing between a child with DS and a child without, as they are given the chance to try for pregnancy again (Asch & Wasserman, 2005). Given that a parent’s actions to ensure an open future for their child seem admirable, the use of selective abortion to ensure an open future for a future child appears to be morally permissible. A similar stance is noted in the writings of Parens and Asch (2000):

[Some feel that] disabling traits are disvaluable insofar as they constrain or limit some opportunities. To say that a disability is disvaluable is only to say that, in the world we now inhabit and in the world we can imagine living in any time soon, to have a given trait would make it impossible or very difficult to engage in some activities that most people would want themselves or their children to have the option of engaging in... traits are disvaluable insofar as they preclude what many find precious. (p. 28)

It may be objected that selective abortion to ensure an open future for a future child comes at the expense of any future for the fetus with DS. However, I am not claiming that a fetus has right; I am claiming that a parent acting in this way is acting consistently with a valid concept of good parenting, and that this is morally permissible.

3.1.7 A fetus with Down syndrome: A lack of potential?

Whether or not a fetus with DS has less potential to live a ‘good life’ than one without DS is difficult to answer. This is, in part, because no single accepted definition of what constitutes a good life exists. Under Glover’s (2006) ‘two-fold’ synthesis of objective and subjective appraisal, a fetus with DS has less potential to live a good life if some requirements, such as health or independence, are required. If these are subsidiary issues, and the ability to love and be loved, to be part of meaningful relationships and keep in close contact with those that matter are of supreme importance, then the fetus with DS would not be disadvantaged. In fact, through some approaches the fetus with DS could have greater potential for a ‘good life’. The judgement depends entirely on certain desirable values, which will differ from individual to individual.

A similar argument applies to the ‘open future’ argument first developed by Feinberg (1980). Having DS precludes some options, such as living independently and
being free of co-morbidities, pursuing higher education and (often) the chance to raise a family. I would argue that it is important for a life to have the option of these pursuits.

I agree that because I value certain experiences this limits my construction of what it might be like not to have these options. I agree that the limitations of DS are not necessarily negative, nor is the absence of limitations necessarily positive. Challenges can be fruitful, helping to develop and mature a human being; many parents describe the ‘challenge’ of raising a child with DS as a positive and rewarding experience (Skotko et al., 2011b). But just because one is presented with a challenge does not mean one must accept it, and many parents will feel that such challenges are best avoided.

Hence, there are two equally permissible attitudes to hold. First, that a child with DS has less potential to lead a good life than one without DS, all else being equal. This is because it can be regarded as a significant extra limitation on the child’s capacity for human flourishing. Second, that a child with DS has a significantly less open future than a child without DS, all else being equal. I support both viewpoints, knowing full well that other conceptions of the good life are equally valid. Pregnant women who are faced with the option of abortion should be free to choose for themselves how a fetus with DS fits into their conception of the good life, too.

On these grounds, I believe that the argument regarding potential holds that a non-DS fetus has greater potential than that of a fetus with DS. Consequently, a fetus with DS has less right to life than a non-DS fetus of the same gestational age.

### 3.1.8 Abortion of a fetus with Down syndrome: Morally permissible, or morally required?

For most women, the decision to abort is probably a choice between raising a child with DS and a non-DS child, as it gives women the option of attempting pregnancy again (Asch & Wasserman, 2005). If we feel that some parents are justified in using selective abortion, does this mean that all parents should abort and try again for another child? The Principle of Procreative Beneficence (PPB) espoused by Julian Savulescu (2001) proposes something similar to this claim. It states that parents have a moral obligation to “select the child... who is expected to have the best life, or at least as good a life as the others, based on the relevant available information.” (Savulescu, 2001, p. 413)
There are several problems with Savulescu’s (2001) command. The first is one which we have already explored; that of ‘the good life’. As long as what constitutes this remains elusive and subjective it will hard to require actions based on this uncertain construct. Savulescu asserts that PPB does not need to answer this question (Savulescu & Kahane, 2009), and that the subjectivity surrounding the good life can be applied by each parent, using their own values and concepts in a way I have described earlier. Through this approach, PPB adds nothing new to our discussion; as we have found, it is a woman’s decision if a fetus with DS will have a less good life than an unaffected fetus.

The second problem is the difference between moral obligations and moral permissibilities. Savulescu (2000) asserts that parents have a moral obligation, instead of a moral permissibility, to select the possible child with the best life. This sets an unusually high standard for parenting, as Buchanan et al. (2000) make clear:

Parents are not morally required to do everything within their power to produce and raise the best children possible. Parents can legitimately give weight to their own interests and to the interests of others besides themselves and their children in making decisions that involve use of their resources or efforts, and in so doing they do not do all that they might do for their children. This would be an unreasonably high standard. (Buchanan et al., 2000, p. 162)

If we accept that parents are not expected to do all in their power for their children, and that they can act to serve their own interests, then the PPB does not imply a moral obligation, but at most a moral permissibility to act in such a way. This is the conclusion which we came to in Chapter 3.1.7.

Savulescu (2009) himself admits that the scope of PPB is limited in relation to other considerations, such as the interests of the parents, existing children, harm to others, and the work involved in the procedure itself. Furthermore, PPB is criticized because of its fundamental reliance on reasons forming a moral obligation, which is usually construed as the other way around; for morality gives us reasons to act in particular ways. This point is well made by Andrew Hotke (2012):

[Using PPB for] any act that we do, if there is some other act that there was more reason to do, then the act that we do is morally wrong. For
instance, if I choose to forgo breakfast rather than eat breakfast, then assuming there was more reason to eat breakfast than not to eat breakfast I have acted morally wrongly; I ought to have eaten breakfast. This conclusion is absurd. (p. 7)

Though we may have acted irrationally when choosing to forgo breakfast, we have not committed a moral wrong. Through Hotke’s (2012) approach, though we may choose a child with less potential for wellbeing we have not acted morally wrongly, but we might be considered to have acted irrationally.

Savulescu (2001) sets a high standard for parenting with the PPB. It seems untenable that PPB implies a moral obligation, and more probable that it implies a morally permissible way of acting. As it answers none of our questions of ‘the good life’, I argue that we hold to our former conclusion; that the abortion of a fetus with DS is more permissible than the abortion of an unaffected fetus.

But are there any fundamental ways in which selective and social abortion are different? Indeed, are there any reasons why a selective abortion could be much more concerning- for parents and for society- than a social abortion?

### 3.2 Selective abortion: Selecting against a trait

For many, social abortions constitute a rejection of parenthood, something to which all women are entitled. This position holds that women have the right to choose when and where they give birth to and raise a child. Selective abortions, however, are enacted on previously well-received, happy and ‘wanted’ pregnancies, where there was no desire for termination. This is because prenatal detection of the selected trait would not normally be provided if the pregnancy was to be aborted anyway.

For social abortions the ultimate aim- a prevention of parenthood- is achieved. For selective abortions however, the same is not true. The previously wanted pregnancy is now rejected on the basis of a trait. The end result- an end of parenthood- is not the anticipated result the woman was hoping for at the beginning of pregnancy.

Adrienne Asch (2000) asserts that a diagnosis of DS instantly turns what is a wanted pregnancy for many women into an unwanted pregnancy with a ‘particular’ trait. In contrast, abortions for social reasons are rejections of ‘any’ fetus; as traits of the
fetus are not known, and indeed even if they were, would have no effect on the desired outcome. The fetus is aborted only because of the circumstances of the mother. From this we arrive at Asch’s ‘any/particular distinction’, where termination of ‘any’ fetus (i.e. social reasons) is acceptable, whereas termination on the basis of a ‘particular’ trait (DS, other medical condition, or sex) is not (Asch, 2000).

This logic is interesting to note, as through it we accept social abortions (for any reason) and reject abortions for any congenital disease, regardless of its severity. This would probably be a controversial judgement to translate into policy, as many who reject abortions for social reasons are prepared to accept abortion for severe disability (Wasserman et al., 2005 p. 4). Testing for anencephaly (where the newborn will die within two weeks of birth) would be rejected under this logic, whereas abortion for an unplanned pregnancy due to disregard for contraception would be permissible. Such a policy ignores the pain and grief which can be placed on parents from such a birth, and overlooks our discourse on assessing moral permissibility in Chapter 3.1.

The justification for this distinction is that rejection of DS expresses and sends a hurtful message to all those with DS, whereas a rejection of ‘any’ fetus does not offend or target any particular group. Of course, this logic asserts that those with DS, and in fact disability in general are a discernible group, a position that has generated much criticism and is discussed in Chapter 4.

### 3.2.1 Critique of the ‘any-particular’ distinction

The any-particular distinction has been scrutinised by several critics, not least of all James Lindemann Nelson (2000). He claims that there is no logical reason that the any-particular distinction should be applied only to selective abortion. He argues that other reproductive decisions could express a similar rejection. Nelson gives the example of a father who chooses a vasectomy, resulting in sterilisation. The parent’s children could feel hurt that the father does not want more children like them, or believe that they are a burden on their father. But the father’s reasons for sterilisation are likely very different; he could feel that he was not justified in bringing more people into an already crowded world, or that he wanted to focus on nurturing the children he already had. Thus, sentiments perceived from any decision may be quite the opposite of the motivations, or presuppositions which generated them. Nelson (2000) believes that the
termination of a pregnancy is no exception; in his words, “abortions are not flags... they
do not wear their meanings on their sleeves” (p. 197).

It is hard not to draw similarities between selective and social abortions when
placed in a real life context. Nelson (2000) uses the scenario of a woman who has
decided that her family of three children is large enough, but is pregnant with another
child. Under Asch’s (2000) framework, this would be a situation of the woman, rather
than a trait of the fetus. However, a quality of the fetus is that the resulting child
would be fourth-born. Does it follow that the woman expresses contempt for fourth-born
children, or large families? In the same manner, does a woman who believes she is too
poor to have a child express contempt for poor children? Nelson asserts that these
relational concepts of poverty, or being fourth-born could equally be the subject of
discrimination, much as DS can be.

Though an interesting argument, these fetal ‘traits’ of being fourth-born or
growing up in poverty are not innate and unchangeable in the way a condition like DS is.
If either of the aforementioned children was put up for adoption the trait of being
fourth-born or poor could be annulled in a way that DS cannot be. Furthermore, these
traits are not so much traits of the fetus but a product of the woman’s situation, as any
child born in these circumstances would be fourth-born or poor (Wasserman & Asch,
2007). They are also substantially different in their stigma and limitations; being fourth-
born or poor does not preclude one’s involvement in society in the same way DS can
(Press, 2000).

Where Asch and Nelson fundamentally differ is the power of the disabled trait in
decision-making, and how much this trait is influenced by stigma and discrimination.
Nelson (2007) makes his view clear:

I expect that many women trying to decide whether to continue a
pregnancy in the face of information about fetal impairments think deeply
about their own hopes concerning parenthood, family life, vocational life,
and so forth, and try to imagine richly how raising a child with the
identified impairment will fit into those aspirations. (p. 475)

Asch’s (2000) view is diametrically opposed to this. She considers a selective
abortion against a fetus with DS as nothing more than a simple rejection of the trait
itself; for “all these claims do not refute the view that [the diagnosis of DS] of the fetus is the basis of the decision not to continue the pregnancy” (Asch, 2000, p. 236).

Asch is correct that prenatal testing for DS only identifies large scale chromosomal alterations and cannot identify anything else. But would we want testing for any other traits? If there became a way to accurately predict the disposition, temperament, or ability of the child to love its parents, would these constitute an adequate reason for abortion? I would suggest that such detection would be a far weaker justification; and as mentioned in Chapter 3.1, the detection of DS is reason enough in itself.

3.2.2 Does selecting against a trait pose any new problems?

It seems probable that for many women a positive test for DS is just another ‘circumstance’ in which women are placed. As a social abortion is a rejection of parenthood, a selective abortion is a rejection of parenthood of a child with DS. It is a rejection of the interplay of the increased challenges that raising a child with DS requires with the woman’s social circumstances. Ultimately, raising a child with DS is a greater challenge than raising a child without DS (all else being equal). Rejecting this does not mean that women devalue or disrespect those with DS.

Thus construed in an individuo-social model, a selective abortion need not present added moral difficulties over that of the social abortion. This position is supported by a substantial body of evidence which shows that the reasons women give for a selective abortion are very similar to that of a social abortion (Kirkman et al., 2009). Thus if social abortion is a morally permissible rejection of parenthood, selective abortion for DS is a morally permissible rejection of what parenthood with DS entails.

But are those who use testing and abortion really making an autonomous decision, or are they acting in accordance with the social pressures of stigma and discrimination? These questions are raised by the synecdoche argument, which is a discussion of the meanings and messages of individual choices for DS testing.

3.3 Stigma and synecdoche

The ‘synecdoche’ objection to prenatal testing has been developed by Adrienne Asch and David Wasserman (2005), though the roots of the argument are explored in
Asch’s early writings on the any-particular distinction (Asch, 2000). The term is derived from its literary use, where synecdoche refers to an occurrence where a ‘part’ stands in for the whole. It therefore asserts that parents allow the trait of DS to fully inform their decision on abortion, though it tells us little about what it is like to raise a child with DS.

The synecdoche argument refers to parents and the reproductive decisions they make; in this case, the act of selective abortion. This argument is concerned with the ideals of family life, and the motives of parents to fulfil these ideals.

Asch and Wasserman (2005) are concerned that a prenatal test only tells parents the presence or absence of DS. It cannot inform the parents of the love the child will have for its parents in due course, or the child’s outgoing capacity in its teens. Since it is a first impression, it is an incomplete impression, and cannot tell us whether raising the child will meet parental expectations. This is similar to other forms of discrimination such as sexism and racism, where a single physical trait is concentrated on, though it tells us little about the individual involved.

For Asch and Wasserman (2005), to test and abort is to undermine a key obligation of parenting: the requirement to show all children “unconditional devotion” (pp. 202-203). By choosing termination, one is unwilling to explore the rest of the individual and the potential which they hold in their life (Parens & Asch, 2000). This view is shared by Mary Johnson (1990):

A decision to abort based on the fact that the child is going to have specific individual characteristics such as mental retardation [sic]... says that those characteristics take precedence over living itself, that they are so important and so negative, that they overpower any positive qualities there might be in being alive. (Johnson, 1990, cited in Asch, 2000, p. 235)

3.3.1 A mistaken approach to the demands of care?

Asch and Wasserman (2005) believe that the extra demands of care that many children with DS require should not be taken into account when considering selective abortion. They detail scenarios where increased parental input is met with joy, in contrast to a diagnosis of DS, which can be met with disappointment and rejection. The diagnosis of a multiple pregnancy is usually well received, despite the increased financial and psychological demands of raising two, three, or even seven children at one
time. Because of this, healthy multiple pregnancies are rarely aborted; when they are, the medical faculty can be shocked (Raymer, 2003).

Similarly, the news that one's child is a musical or mathematical ‘child prodigy’ is typically met with excitement and celebration, despite the added parental demands that nurturing these qualities will entail. Asch and Wasserman (2005) ask why these changes in ability are welcomed, whereas the added demands for a child with DS are accepted with reluctance.

Asch and Wasserman (2005) conclude that it is not the increase in psychological or financial demands that is rejected by prospective parents. Rather, it is the fact that these traits- or more specifically, individuals with these traits- are not stigmatized against. Twins and triplets are not a stigmatized group, and neither are musical prodigies; because of this, the increase in parental caregiving is welcomed as a privilege. Repeated maths exercises and expensive schooling are not stigmatized against in the same way that repeated visits to the doctor are. In contrast the label of ‘DS’ has stigma attached, and this is the reason that parents may see such occurrences as a ‘burden’.

Though Asch and Wasserman (2005) assert that stigma is the only reason as to why a child with DS can be seen as a burden, I would suggest that there are other issues at play. Investing time in a musical prodigy is not required in the same way that increased care for children with DS is; for parents it is a choice, not an obligation, whether or not to develop a musical talent. It is true that twins and triplets require more parental time and effort, but they reach a stage of independence far quicker than children with DS (who are likely to retain some dependency throughout life). Though repeated maths exercises or sports practices may be time-consuming and wearisome for parents they are (for the most part) allowing a child to grow or fulfil a passion. In contrast, repeated hospital visits are not so much about facilitating growth as preventing decline, along with the anxiety and disruption that these events can cause.

Raising a child with DS is not identical to raising children with non-medical needs. They require increased levels of support, and may require greater levels of medical care. These requirements are significantly different to the voluntary facilitation of a specific talent, or the upbringing of multiple offspring. Because parents may act in their own interests, that children with DS place significant demands on caregiving is yet
another reason why it is more permissible to abort due to DS, as opposed to ‘any’ reason.

Though this may be true, Asch and Wasserman (2005) would suggest that stigma and discrimination are still influencing this judgement; they are making the upbringing of a child with DS seem more demanding and less desirable than it really is. Thus it is pertinent to ask: how prevalent is stigma in today’s society, and in our day-to-day thinking?

3.3.2 Stigma: Both prevalent and pervasive?

Asch and Wasserman (2005) assert that reproductive decisions surrounding prenatal testing are driven not by parents pursuing admirable parental goals, but by mothers and fathers who allow the social pressures of stigma and discrimination to dictate their choices. For our purposes, such a claim is premised upon stigma and discrimination existing towards those with DS. What is the evidence for this?

The literature suggests that some discrimination towards people with DS and intellectual disability exists. Members of the public often show a limited understanding of intellectual disability (Gordon et al., 2004), and that they often underestimate the prevalence of intellectual disabilities (Tachibana & Watanabe, 2004a, 2004b). People with intellectual disabilities are often one of the least desirable groups to interact with in social situations (Gordon et al., 2004; Nagata, 2007; Westbrook et al., 1993). Attitudes and intentions are often more negative towards those with intellectual rather than physical disabilities (Brown et al., 2011). These negative attitudes have been partly linked to the public misconception of intellectual disability, overestimating the severity of the impairment on the affected person’s life (McCaughey & Strohmer, 2005). These findings indicate that public knowledge and acceptance of individuals with intellectual disabilities, (of which DS form a part) is limited.

That some stigma remains against people with intellectual disability is undeniable. However, this does not mean that all women have severely negative attitudes towards DS, such that it would be the only significant factor in decision-making and thereby eclipsing the whole experience of raising a child.

Data in the literature supports this viewpoint. In developed countries there is widespread support for the integration of those with intellectual disabilities into society
Although some oppose the integration of the cognitively impaired, this number is small and may not necessarily harbour negative sentiments but be a (misguided) envisioning of the environment in which people with disabilities thrive (Scior, 2011; Tachibana & Watanabe, 2004a). A number of studies have indicated that females are more likely to report positive attitudes towards intellectual disability than males (Gash et al., 2000; Nabors & Keyes, 1995; Nowicki, 2003), though some studies have detected no difference (Tamm & Prellwitz, 2002). Younger people and those with higher educational qualifications also tend to express more positive attitudes (Akrami et al., 2006; Burge et al., 2007; Macdonald & MacIntyre, 1999). Though evidence is scarce, a number of longitudinal studies suggest that there has been a positive shift in attitudes towards the intellectually disabled over time (Rees et al., 1991; Tachibana & Watanabe, 2004a).

These positive findings are echoed by several studies which have looked solely at attitudes towards those with DS. Though some people have inaccurate knowledge of the causes of DS, the majority of adults and youths favour the inclusion of children with DS in the classroom (Gilmore et al., 2003; Pace et al., 2010). The majority of adults feel that those with DS are employable (Pace, 2011). Similar to attitudes towards generalised intellectual disability, female sex and higher education are related to more positive attitudes towards people with DS (Pace et al., 2010). One study found that 85% of teachers and 78% of the community regard the defining feature of a child with DS as positive, such as affectionate, happy or friendly (Gilmore et al., 2003). In another study, nearly all participants labelled those with DS as ‘affectionate’ (Wishart & Manning, 2007). This study also indicated that most had optimistic predictions for milestones (such as independence, employment and marriage) that people with DS could achieve.

Stigma persuades someone to act in a particular way. Viewed in this way, stigma is just another (albeit bad) reason for selective abortion. It does not make selective abortion in itself wrong, but may give rise to an irrational decision for the woman involved. In short, that stigma can be implicated in the decision of selective abortion does not make abortion for DS any less permissible.

With this in mind, it is likely that the power stigma holds over individual reproductive decisions is limited, and is at most just another factor implicated when considering pregnancy termination. In the same way that the any-particular distinction
was rejected for its denial of the plurality of women’s reproductive choices, it is likely that social stigma plays a role which is limited in a woman’s consideration of selective abortion.

3.3.3 Does prenatal testing and selective abortion devalue the process of parenthood?

Regardless of the role of stigma, Asch and Wasserman (2005) assert that a diagnosis of a condition such as DS is not a good predictor of the future family life and the fulfilment of parental goals. Their versions of permissible goals include: being able to give and receive love, and to nurture and raise a child with ‘unconditional devotion’. These are non-specific, abstract parental goals that can be met just as easily, Asch and Wasserman argue, by raising a child with DS, or raising a child with the loss of a limb, or raising a child who is ‘normal’, or one that has abnormally high intelligence; the traits themselves are unimportant. It is clear that some specific parental goals may not be met by a child with DS; such as excelling at a certain sport, or pursuing a certain career (a child with DS will not become a doctor like her mother). However, Asch and Wasserman argue that specific goals are not the goals which parents should have. To have specific goals is to be ‘consumerist’ in a moderate sense (vague parental goals) and ‘perfectionist’ in an extreme sense (exact parental goals).

Asch and Wasserman (2005) assert that the uniqueness of any child should be praised; not measured by comparison to any defined ‘norm’. Those with DS can love and be loved, can act selflessly and with thought, can reflect on the world around them and can contribute to the community. Asch and Wasserman assert that these are the things that matter in a child, and that this devotion should begin before birth. In short, parenting should begin with unconditional welcome.

For Asch and Wasserman (2005), families represent a distinct and valued form of association where no selection exists. They assert that reproductive technology continues to erode the concept of an all-accepting family, making it more like an exclusive “club” where acceptance is contingent on “perfect health” (Asch, 2000, p. 239). If prenatal testing enforces the idea of required ‘acceptance’ into the family ‘club’ then, in turn, will members of the family be rejected when they develop disabilities in their old age? Will children be rejected, if they develop a debilitating illness or accident in their youth? Asch and Wasserman believe that parental attitudes and expectations
should undergo a paradigm shift; from one that is focused and specific, to one which is uncritical and accepting.

Asch and Wasserman’s (2005) concept of unconditional welcome may be helpful for parents, as all children inevitably diverge from parental preferences, and raising any child contains some form of uncertainty. But it is also strict, asserting that parenthood should begin once the choice to bear a child is made. This is far before the generally recognised beginning of parenthood, which is usually childbirth. If parenting exists in pregnancy, does this mean that the significant numbers of families who have used prenatal testing (or more pertinently, aborted a fetus because of this) are not welcoming? If they are not unconditionally welcoming at the pregnant stage, does this mean that they are ‘bad parents’ to their children? Do those who reject prenatal testing necessarily become ‘good parents’?

At most, unconditional welcome could be an admirable value. But requiring all parents to show unconditional welcome sets an unacceptably high standard for parenting, for some congenital diseases cause much pain and anguish. Must all parents suffer these harms because the pregnancy was previously wanted, only to have their child die within a number of weeks? It seems only fair to give women a choice, as forcing this on parents seems unnecessarily harmful. Parenting is not a solely selfless act, and parents have their own interests which should be taken into account in any reproductive decision. In the same way that Savulescu’s (2001) Principle of Procreative Beneficence is more a moral permissibility than a moral obligation, Asch and Wasserman’ (2005) value of unconditional welcome may be an admirable approach to raise a child, but it is not a philosophy which must be adhered to.

To reiterate, Asch and Wasserman’s (2005) appropriate parental goals include being able to give and receive love, and to nurture and raise a child with unconditional welcome. Specific goals, such as raising a child to become a doctor, should be eschewed. However, there seems to be a grey area between goals which seem ‘appropriate’ and goals which seem ‘unnecessary’. What of the ‘goal’ in raising a child which has, within reason, the potential to follow whatsoever path he or she desires? Wanting a child to have the option of pursuing a life in academia is similar to wanting the child to have the option of pursuing sports, or music. One can accept the child pursuing all, or a selection, or none of these, and support the child nonetheless. Is aiming to raise a child which has a
reasonable array of these options morally repugnant? By acting to ensure Feinberg’s (1980) ‘open future’ we can still comply with Asch and Wasserman’s need for unconditional acceptance and support, though we may not accept a fetus with DS.

3.3.4 Is the synecdoche argument convincing?

The synecdoche argument is useful in identifying the role of discrimination and stigma in the attitudes and motives behind parental decision-making. Although some could argue that this is reason for the cessation of prenatal testing, it would seem that the most obvious way forward is to improve the education of those who receive screening and diagnosis. This education would involve materials which emphasised how all people with DS differ in their individual characteristics, with accurate descriptions of a life with DS. These materials would help to minimize the role of stigma in parental decision-making, ensuring that ‘the part’ does not stand in for ‘the whole’.

Asch and Wasserman (2005) concede that their demand for unconditional acceptance which begins from conception is a high, idealistic standard for parenthood and may not be realistic (Wasserman & Asch, 2007). Nelson (2007) is particularly sceptical of this value, feeling that ‘unconditional welcome’ runs counterpoint to the direction most parents encourage their children to take, a path which is nonetheless still consistent with good parenting:

We mold [sic] our children right from the start, incorporating them into particular regimens of practice, evaluative outlooks, and physical habits. If our children are ill or disabled, we do not accept such states of affairs as expressing our welcome of them ‘as they are,’ and it is a good thing, too. Rather, we subject our kids to considerable inconvenience and pain to enculturate, to cure, or to compensate. (Nelson, 2007, p. 478)

Wasserman and Asch (2007) maintain that the idea of unconditional acceptance is still consistent with directive parenting. Nevertheless, there is no way in which unconditional acceptance or welcome should be considered a moral obligation. As the ideals of parenthood are subjective, contested, and limited in regards to other interests, I assert that the synecdoche argument is no more than another consideration with which to examine parental motives for testing. There is little to indicate that by preventing women from accessing testing we would increase parents’ feelings of unconditional
acceptance, or encourage better parenting; for it follows that one should treat the cause of discrimination, not the effect.

### 3.4 Conclusion

Abortion is now available to most of the world’s women. For many of these women, social reasons are deemed suitable to justify abortion. Many feel that the presence of significant impairment in the fetus is also sufficient reason for abortion. Indeed, though some objectors may oppose abortion in principle, most believe that abortion in the case of significant impairment is permissible.

This chapter aimed to find what might make a selective abortion for DS more morally permissible than a social abortion for ‘any’ reason. It is usually argued that because many conditions demand increased parental care that this can be a moral ‘harm’, and therefore selective abortion is more justified (Shakespeare, 2006). Though I agree that this is a relevant consideration, this may not be the only one.

My approach to this question has been original, by applying recognised arguments for fetal ‘right to life’ specifically to the fetus with or without DS, and by re-casting the argument for potential in a new light. Viability and sentience perspectives were relatively inconclusive, indicating little difference in right to life between a fetus with DS and an unaffected fetus. However, an examination of the fetus’ potential to live a good life and have an open future indicates that the potential of a fetus with DS is diminished (though it is by no means non-existent).

Not all women will feel this way; some will believe that people with DS are not restricted in the options which matter. This is a valid perspective to have, as what constitutes a good life is subjective and contested. For these reasons, it is more morally permissible to abort a fetus with DS than to abort a fetus without DS. Applying a similar framework of enquiry to the viability, sentience and ‘potential’ criteria to other disorders may reach similar conclusions.

It should be noted that some social reasons for abortion are especially strong, such as in cases of rape or incest. However, for most other social reasons, the previous discussion indicates that selective abortions are more permissible. My argument is
premised on abortion being justified up until 20-24 weeks of pregnancy. If this is incorrect, then my argument is limited.

That selective abortion for DS is morally permissible does not mean that it is morally praiseworthy, nor does it mean it is a moral obligation. Selective abortions for DS involve a rejection of parenthood of a child with DS, and are not a direct rejection of people with DS. Stigma and poor parental goals may influence some reproductive decisions, but the right approach is to confront the reasons behind these motives, not by banning one of their outlets. At present, discrimination and stigma towards DS is not as pervasive as Asch and Wasserman (2005) might assert, and there is some evidence of improvement.

Consumerist and perfectionist approaches to parenting are particularly troubling, as they are likely to limit the child’s open future. But abortion in the case of DS is not an example of consumerism or perfectionism. If a woman is aware of the stigma surrounding disability, and accurately informed what lives are like with DS, then this should not make her choice to abort a fetus with DS unethical. If we regard abortions for social reasons as an acceptable expression of reproductive autonomy, then we regard abortions for DS as reasonable, too.

What, then, of selective abortions for minor diseases such as asthma, or eye colour, or sex? Could these not preclude certain options; for example, being a woman in India? As these traits constitute only minor limitations, they do not significantly impact on the good life. All children with these traits still have an open future. Caring for children with these conditions significantly disrupts parents’ lives. Abortion constitutes a significant and value-negative deed, and a woman’s reproductive autonomy is limited in regards to these and other considerations. Thus a fetus with any of the above conditions would have a right to life similar to that of a non-DS fetus, and would need the likelihood of a significant current or future harm to justify abortion. In contrast, a fetus with a severe impairment like anencephaly (where the brain is exposed at birth and the newborn dies within the first two weeks of life) would have less right to life than a fetus with DS, and it would be more morally permissible to perform an abortion.
Chapter 4 MESSAGES AND MEANINGS: THE OFFER OF TESTING AND ABORTION FOR DOWN SYNDROME

In the previous chapter, we arrived at the conclusion that selective abortion in the case of DS is morally permissible, and more permissible than that of a social abortion. The primary aim of prenatal testing for DS is to allow women abortion in the case of a positive result. It is indeed true that a small number of women choose invasive diagnosis only to prepare for the birth of a child with DS, with no intention of aborting a DS-positive fetus (Antenatal Down Syndrome Advisory Group, 2007). However, the majority of women choose testing to allow the option of abortion, shown in part by the large proportion choosing to abort in the case of a positive result (Mansfield, 1999; Natoli, 2012).

Testing is contingent on the permissibility of selective abortion; if selective abortion was morally objectionable then allowing the option of it via testing would be morally repugnant. Because selective abortion for DS is permissible, it can be argued that an agent which allows it to be carried out is *prima facie* ‘value-neutral’, and also permissible. The agent in this case is prenatal testing. As testing is permissible it may be made available, thus it may be chosen by some women. As long as there is a demand for testing, research and development will continue to strive to meet this demand.

However, some authors contend that the availability of prenatal testing is *not* value-neutral, and that the offering and use of testing and abortion ‘express’ underlying prejudices or other modes of thought which are hurtful (Parens & Asch, 2000). This stance on prenatal testing has been labelled the ‘expressivist’ position, and encompasses much of the objection to prenatal testing by disability rights activists and others. Some commentators have gone so far as to claim that “prenatal [testing] and selective abortion communicate that disability is so terrible it warrants not being alive” (Asch, 1989a, p. 319).

The *individual* uses of testing and abortion and their capacity to reflect a particular rejection or stigma were dealt with in Chapter Three. This chapter aims to analyse the capacity of the *collective* use of prenatal testing and selective abortion for DS to express discriminatory views on disability. To do this, the logic of the expressivist position must be discussed.
Little weight is given in this chapter to whether research and development of tests is discriminatory. Instead, the focus is the possibility of the more ‘hurtful’ availability, offer and use of testing and selective abortion. This is because research and development of prenatal tests are predominately driven by demand. These actions by themselves do not create discrimination; at most, they reflect it. This is in contrast to the view of some disability rights positions (Asch, 2000), though it does not make up a large part of their objection.

4.1 Does testing for Down syndrome express hurtful attitudes?

The expressivist position is a common argument against the use of any form of genetic selection, and so opposes not only prenatal testing and selective abortion, but also preimplantation genetic diagnosis and gamete selection processes as well. This general group of concerns towards the ‘messages’ of genetic selection was first labelled as ‘expressivist’ by Allen Buchanan (1996):

The expressivist objection... focuses on what may be called the expressive character of decisions to use genetic interventions to prevent or remove disabilities... The claim is that the commitment to developing modes of intervention to correct, ameliorate, or prevent genetic defects expresses (and presupposes) negative, extremely damaging judgements about the value of disabled persons. (p. 28)

Under this framework, prenatal testing for DS is exactly the kind of intervention which expresses these ‘hurtful attitudes’. These harmful judgements can be reflected in both the choices of women regarding testing, and the framework of testing and selective abortion itself. According to Buchanan (1996), an example of such a received judgement would be that “the lives of individuals with disabilities are not worth living” (p. 28). Similar objections have been raised by disability activists such as Marsha Saxton (1998):

The message at the heart of widespread selective abortion on the basis of prenatal diagnosis is the greatest insult: some of us are ‘too flawed’ in our very DNA to exist; we are unworthy of being born... [F]ighting for this issue, our right and worthiness of being born, is the most fundamental challenge to disability oppression; it underpins our most basic claim to
justice and equality- we are indeed worthy of being born, worth the help and expense, and we know it! (p. 391)

These messages are ‘societal’; that is, they can be received and interpreted by everyone. Some may regard these messages as indicative of successful parental choice in pregnancy. Some may regard prenatal testing as a useful method to reduce the prevalence of disability. For these people, the messages ‘sent’ by prenatal testing are therefore construed positively. Others will see these messages in a negative light, perhaps because they feel prenatal testing undermines the lives of those with disabilities. They may feel that these negative messages portray negative judgements, and therefore prenatal testing precludes the full integration of people with DS into society.

By extending the concept of the any-particular distinction, Asch and Wasserman (2005) assert that it is logical to perceive offence when a particular trait is the basis for the prevention of a possible life, and that trait is shared by a group which has been stigmatized throughout history. They further contend that the substantial time and effort to obtain results together with the traumatic and painful action of abortion all show the lengths parents and medical professionals will go to, to prevent the birth of another ‘like them’.

There are both ‘consequentialist’ and ‘non-consequentialist’ approaches to the expressivist argument (Shakespeare, 2006). A consequentialist approach states that these ‘messages’ result in real consequences- i.e., actual harm. Such a harm might include increasing societal stigma to disability, further marginalising those with DS through discriminatory attitudes. Those who espouse the consequentialist expressivist position assert that prenatal testing for DS violates and jeopardises the full inclusion and acceptance of people with disabilities in society. This is problematic, as it is often the social barriers to participation and inclusion which make up a significant portion of the burden of disability (World Health Organisation, 2011). Therefore if the consequentialist expressivist argument is valid, prenatal testing is making the lives of those with DS and other disabilities more difficult.

Non-consequentialist approaches do not state that actual harms eventuate from the practice of testing and abortion; yet concern remains over prenatal testing due to the mere fact that discrimination can be perceived by such practices. Both approaches to the
expressivist position conclude that the sentiments giving rise to prenatal testing are hurtful. Parties particularly likely to interpret prenatal testing and selective abortion for DS as hurtful could include:

1. Most directly and most importantly, living individuals with DS.
2. Those with congenital cognitive disability (e.g. spina bifida, fragile X syndrome).
3. Those with congenital physical disability (e.g. cystic fibrosis, achondroplasia).
4. Those with acquired cognitive or physical disability (e.g. brain injury from a motor vehicle accident).
5. Those concerned with the rights of the disabled, including family, friends and caregivers of groups 1-4.

4.1.1 Critiquing the reasoning of expressivism: The fallacy of one-dimensional messages

Although various messages can be derived from an action, it does not follow that any message is necessarily correct. For example, because I always have sugar in my tea does not mean that I support a far-right political party; such a message to perceive from such an action would be unreasonable. Therefore we must assess the legitimacy of the expressivist claim that it is logical to perceive a harmful judgement from testing and abortion for DS.

A strong criticism of the expressivist argument is that it is impossible to conclude the exact ‘message’ that is created by any woman’s voluntary decision to undergo testing. This idea is one which has been used to defend the tenets of virtue ethics; that it is careless to attribute a virtue to a person on the basis of one or more actions, especially if the motive for the person’s action is unknown (Sreenivasan, 2002). Acts, and the messages they convey, rarely have a single motivation or meaning. As Weinstock (2010) has written, “it is a perilous exercise indeed to impute a univocal meaning to an action... that people doubtless undertake for a variety of motives” (p. 224).

Though some prospective parents may have wholly negative attitudes towards people with DS and this may be the only reason to influence a decision, it is reasonable to assume that most choices will be driven by other factors. For example, a family may already have a child with DS, and feel that they do not have the emotional or financial
capabilities to give another child with DS a good quality of life. The mother of another family could have a disability themselves, and feel that this is something she would like to prevent in her children. In these scenarios, the choice to test and/or abort is probably not born out of negative sentiment or discriminatory attitudes, and is more likely an accurate understanding of the demands of a child with DS and the limitations of the family. Indeed, many groups which advocate for widespread prenatal testing include families with children who have the disorder in question (Weinstock, 2010).

In many cases, any negative messages are extremely heterogeneous, as are the circumstances and motives that give rise to them. Perhaps, then, to receive a hurtful message is to assume all choices for testing or selective abortion are born out of discrimination and negative sentiment:

[the disabled] cannot, in other words, point to the discriminatory attitudes or statements of others to show that they are being harmed, in the way, say, that a Jewish person might be able to point to an anti-Semitic statement as in and of itself constituting harm [as there are many attitudes conveyed in the use of prenatal testing] (Weinstock, 2010, p. 224)

The argument against the expressivist argument is thus similar to that against the any-particular distinction: for it is inappropriate to attribute a single motive to a pluralistic action. Indeed, any motive or message conveyed by testing and abortion is likely to be misinterpreted, as no message has been purposely created; these choices are made by prospective parents who are acting to fulfil their own familial goals (Parens & Asch, 2000). This, however, does not stop people with disabilities feeling harmed by such an act. For whether or not a message is intended, many still feel disrespected by prenatal testing.

4.1.2 Expressivism developed: The process of testing and abortion

Much of the critique of expressivism focuses on the heterogeneity of messages. It states that it is illogical to conclude that individual choices for prenatal testing and selective abortion necessarily reflect discriminatory attitudes. However, the expressivist objection is not only aimed at the individual choices of women, but also the permission of
testing and selective abortion by the state, and the *offering* of testing by health professionals in a therapeutic context.

These processes are independent of reproductive choices by parents; and it is to these processes that the expressivist argument has more recently been applied (Asch & Wasserman, 2005). Aimed at the framework and language of testing and abortion, the expressivist argument is directed at the concept of choice; namely, that the views of health professionals and governing bodies are so negative and pervasive that choice is constrained and directed towards termination. Reproductive autonomy is *limited, not enhanced* by testing. Thus, women cannot choose freely and their decisions will not reflect discrimination, but more a forced participation.

Asch and Wasserman (2005) assert that it is the systematic, organised prevention of disability by testing and abortion which offend those with disability. However, the prevention of accidents, disorders and disease takes place every day, on a wide scale. How, then, is the prevention of DS different to the prevention of the common cold?

### 4.2 Expressivism: Opposing selection, opposing prevention?

[Prenatal testing] targets unborn children with Down syndrome for selective abortion... persecuting persons with Down syndrome through the prevention of their births. (Saving Downs, 2011a)

This position, held by Saving Downs (2011a), a New Zealand-based anti-testing group, is a form of the expressivist position. It states that the prevention of a disorder like DS necessarily offends people with DS.

A major criticism of this position centres on its logical expansion; namely, that the prevention of other diseases is equally as discriminatory. How, then, is the prevention of DS morally different to the prevention of disability and illness in other areas of society?

Bonnie Steinbock (2000) has argued that all reductions in disease incidence by prevention are morally equivalent. We seek to prevent disability constantly in our community; for example, the use of high-visibility clothing in roadwork and construction. Do workplace safety laws which reduce the number of disabled persons
offend the disability community too? And if not, why not? How does this compare to preventing flu by a flu vaccine, or preventing food poisoning by using latex gloves to prepare food? Perhaps these all differ from prenatal testing as they are prevention of disability and maintenance of health to existing human lives; more adequate examples, then, would be related to reproduction and pregnancy.

Most pregnant women take great care in their diet and lifestyle over the course of gestation, to ensure the health of the resulting baby. Alcohol intake is limited for many reasons, not least of all to prevent Fetal Alcohol Syndrome in the developing fetus. Spina bifida results in varying levels of intellectual disability, and is caused by the failure of the neural tube to close during development. It has been shown that the incidence of this can be vastly reduced with folic acid supplementation around the time of conception (Honein et al., 2001). Cigarette smoking is discouraged in pregnancy, and has been linked to poor birth outcomes such as miscarriage, stillbirth, prematurity, and low birth-weight (Steinbock, 2000). These low weight, premature babies are at increased risk of developing neurological problems.

Interventions such as a cessation of smoking, limiting of alcohol, and folic acid supplementation around the time of conception are almost universally accepted as responsible behaviours, because they prevent disability and poor birth outcomes in the neonate (Steinbock, 2000). These measures are strongly encouraged in many countries, and not partaking in them can be seen as reckless.

Both the routine medical care described above, and the selective abortion of a child can be described as types of ‘prevention’. For some commentators, it is reasonable for parents to go to lengths to prevent an outcome that they may reasonably want to avoid (Steinbock, 2000). However, I believe that there are two integral differences between these routine aspects of prenatal care and the offer of prenatal testing.

The first difference pertains to the outcomes of the test. Routine prenatal care aims to maintain the health of the fetus, whereas prenatal testing for untreatable congenital conditions is only of benefit to the parents. The second difference is the action required by the woman. The fortification of a woman’s diet of a certain amount of folic acid, or a restriction on consumption of alcohol is not a major life change. In contrast, the termination of a pregnancy is psychologically far more disturbing, and requires a much larger motive. Because this ‘motive leap’ is so huge, it shows the lengths
that women will go to, to prevent disability in their child; making the expressivist ‘message’ that much more potent. As Asch and Gellner (1996) have written, “[w]hat differentiates preventing disability by abortion from preventing it by immunisation is that the abortion indicates that the disability makes the child unacceptable” (p. 339).

Abortion is not simply the prevention of disability; it is the prevention of a human life. It is the magnitude of this act which may indicate that a life with disability is unacceptable; in contrast with the act of immunisation, which is a relatively painless and quick procedure. Though immunisations, the use of seatbelts, and the avoidance of certain foods in pregnancy can still be considered as expressivist, we can conclude that testing for DS is suitably different to many other actions taken to avoid impairment.

But still, the question remains: how can people with DS and others logically take offence? This requires a link between DS and a person’s identity which is different to that of many other conditions. It is to this subject which we now turn.

### 4.3 The necessary link between expressivism and identity

Some argue that because prenatal testing results in the prevention of DS, those with DS are being discriminated against (Saving Downs, 2011a). This argument depends on the premise that part of one’s identity is shaped by the trait which is being tested for; if “disabled persons are their disabilities” (Baily, 2000, p. 65) in an ‘identity-defining’ way. This makes prevention via prenatal testing different to the treatment and prevention of other impairments or illnesses, which scholars such as Edwards (2004) regard as non-identity-defining:

> Just as it would be implausible to suppose reduction of the incidence of illnesses such as flu sends a negative message to ill people, so it is not plausible to suppose prevention of disability sends a negative message to disabled people. The expressivist objection hinges, however, upon a view of the relationship between disability and self identity which sees disability as part of the identity of the disabled person, in a way in which illnesses such as flu cannot be. (p. 418)

What, then, does one mean by ‘self-identity’? Many philosophers, from Aristotle to Derek Parfit have attempted to define exactly what is meant by the ‘self’. Personal or
self-identity can be defined as ‘that which makes a person that person’. Some have defined personal identity as a “series of qualities that make it possible to identify the individual as belonging to a particular type or kind or class of individuals” (Glas, 2006, p. 133). For example, my height, the colour of my eyes, the way I speak and my love of sports are all qualities which make me ‘me’.

Others have developed a more relational view of personal identity. Grant Gillett (2008) asserts that “[a] human individual is located in a network of relationships in which he or she is given a name and develops an identity through that being-in-the-world-with-others, an engagement normally contingent on a certain (kin-based) origin” (p. 51). This relational view holds that personal identity develops through the interaction of three key aspects: biological origin (genetic make-up), psychological integrity (consistent mind-narrative through time) and environment (social and physical interaction). Though this is a broad definition it is necessarily so, because identity is understood as a complex formation of many parts, and is not reducible to any singular entity.

We will accept Gillett’s (2008) criteria for assessing what constitutes self-identity, and will look at applying it to certain situations. Through this we can address Edwards’ (2004) challenge of whether DS is ‘identity-defining’ in a way that other disabilities such as Alzheimer’s disease, or other illnesses such as ‘the flu’, are not.

4.3.1 Identity-defining disability: The case for Down syndrome

Gillett (2008) asserts that one’s biological origin- their genetic make-up- contributes to a person’s identity. This seems intuitive; for many things which make me ‘me’ have been influenced by my genes; e.g. the shape of my body, the colour of my hair and the restriction of my vision. Accepting that one’s genetic makeup is important does not mean that we embrace genetic essentialism, as psychological and environmental factors also play a part in the development of identity. For example, one way in which the environment acts on genetics is the non-expression of some traits due to insufficient nutrition or through poor upbringing.

While it is obvious that all with DS share a significant biological origin in the form of three 21st chromosomes, I would argue that it is not the individual nucleotide or chromosomal arrangement which defines our identity but the traits expressed by it
which are important. For example, I know nothing about the arrangement of base pairs on my Y chromosome, yet I know that I am male. For the same reason I would argue that those with DS do not identify with their three 21st chromosomes, but with the traits expressed by these.

The biological traits shared by all with DS number only two. These are similar (but not identical) phenotypic features (particularly in the facial region), and similar (but not identical) levels of cognitive impairment. Other co-morbidities vary in their occurrence and are not shared by all people with DS. Cognitive impairment and similar phenotypic features are therefore the only two predetermined parts of 'biological origin' which are relevant for this discussion.

Both of these traits are present from birth. A person’s self-understanding begins from, or soon after, birth. Self-understanding contributes to one’s personal identity; for who I think I am is decided by what I understand myself to be. In the same way, personal identity forms gradually throughout life, within the nexus of relationships of which we are all part (Gillett, 2008). How we interact with others depends in part on one’s (genetically predetermined) traits. Interaction with a cognitively impaired child will be different to how we interact with a non-impaired child; interaction with a cognitively impaired adult will be different to that of a non-impaired adult. The individual with DS will progress through life and form their identity in a significantly different way to that of the non-DS individual, because the two traits in question are ever-present.

Thus, DS would contribute to self-identity for two reasons: because the condition is present from birth and because it significantly alters interaction with one’s social and physical surroundings. The condition’s cognitive limitations do not alter the remaining criterion of psychological integrity, but may affect the way a person with DS understands themselves to be.

While DS contributes to identity, it seems an overstatement to label the condition ‘identity-defining’, as it seems to suggest that all people with DS are very similar. This approach could be accused of being synecdochal, stigmatizing and inaccurate. Those with DS are individuals, and variable like all others. Thus I will use the term *identity-forming*, suggesting that for people with DS this is only a part of their personal identity.
4.3.2 Are all disorders identity-forming?

Does this mean that all congenital conditions are identity-forming? Fragile X Syndrome and spina bifida are other examples of congenital conditions which often result in moderate cognitive impairment, and it seems reasonable to conclude that both would be identity-forming in a way similar to DS. A condition like mild asthma would not be, as it will not influence events significantly through interaction with the physical or social environment. But what of phocomelia, a congenital condition which results in one or more limbs being unable to develop? The condition is present throughout life, but as the limitations imposed are purely physical it seems plausible to argue that the social interaction or “being-in-the-world-with-others” (Gillett, 2008) need not be altered in such a significant way. Though phocomelia is identity-forming, it is not as much as DS might be, under Gillett’s (2008) broad approach to identity.

Thus it seems that cognitive impairment influences one’s life in a more identity-forming way than physical impairment. This seems reasonable, for the ability to interpret the world around us, reflect on one’s life and interact with others is heavily influenced by cognitive ability. This is not to say that cognitive ability is the only contributor to one’s identity— one defines oneself by one’s actions, deeds and body image, etc. It is probable that the greater the physical impairment the more identity-forming it will be. But I suggest that cognitive ability is notably different as it significantly influences ones’ understanding of self and interaction with the world around us.

What, then, of acquired disabilities; specifically ones where cognitive ability is impaired? Can these be identity-forming too, and why might we not hesitate to prevent the occurrence of such disabilities? We strive to prevent debilitating illness throughout society; the use of hard hats in construction zones, and the medical assessment of concussed rugby players are but two examples. We attempt to treat intellectual disability; consider the case of Alzheimer’s disease (AD), which results in memory loss and progressive cognitive impairment:

A treatment for Alzheimer’s disease would result in a society where ‘different’ people existed than one where Alzheimer’s is not cured. If this is true, then attempts to eliminate Alzheimer’s may well carry an implicit statement devaluing or rejecting those affected by the disease. Can we
therefore conclude that society devalues or disrespects or rejects those with late-stage Alzheimer’s? (Gavaghan, 2007, p. 27)

In this passage, Colin Gavaghan (2007) asserts that the progression of AD increasingly changes one’s identity. This is understandable; AD patients are certainly different to how they were before disease onset. Having AD will considerably alter social interaction and one’s network of relationships over a course of time, such that the person one or two years since the initiation of the disease may be very different to the person before the disease began. Indeed, the psychological integrity of the person has been jeopardised, with the memory loss from dementia contributing to a loss of sense of identity. Using Gillett’s (2008) conception of personal identity, the identity of the AD patient has changed.

If we strive to cure Alzheimer’s, are we not removing ‘part’ of the identity when we remove this disease? Would we necessarily devalue those with AD when we treat them, and more so with a cure? With treatment, do we result in ‘changed’ persons? One could argue that AD is a progressive, acquired disability- one which morphs from an already-established identity. Treatment aimed at Alzheimer’s aims to restore the prior self. I propose that AD is an identity-morphing illness, one which is not integral to one’s identity but a pathological feature which becomes intertwined with identity over time. Acting to prevent and treat AD is different to preventing DS because DS is a congenital condition which the identity has developed with, not deviated from. Similar arguments could be made to support the treatment of identity-morphing psychiatric conditions, such as borderline personality disorder (BPD).

But if acquired conditions such as AD and BPD can be partially identity-forming, where does the distinction end? Contrary to Edwards’ (2004) statement, is the flu identity-forming as well? Having the flu alters social interaction, and can result in some degree of psychological disturbance. People with the flu are labelled as ‘having the flu’, aiding diagnosis, management and allocation of resources (in the same way that ‘labels’ are an inescapable part of any illness or disability). Why, then, might acquired conditions like the flu be less identity-forming than something like AD?

The most significant reason is that influenza is a transient illness, and the limitations conferred by it have an expected end. One’s identity is not formed separate in any instant in time, as we remain the same person from moment to moment. Thus, self-
identity requires some measure of narrative continuity. The flu will not end this narrative continuity, unless in the case of death. But the flu will have some bearing on the social environment of the patient; it may limit or strain interactions with others, and others will change their actions accordingly. This suggests some minor identity ‘morphing’, under Gillett’s (2008) approach towards identity.

So it seems plausible that at the time of illness the flu is only minimally ‘identity-forming’, if at all. What, then, of acquired conditions which cannot be cured, such as diabetes, or hypertension? How can we act to prevent and treat these conditions while not offending those with the condition? The answer to this is the same as that for AD: with all acquired conditions, a limitation is imposed on an already-formed identity. These conditions are ‘identity-restricting’, through our definition of personal identity which encompasses social interaction. These chronic conditions are not a part of the already conceived ‘self’. Therefore, treatment, is an identity-preserving intervention (Hope & McMillan, 2012). Workplace safety laws, and polio vaccinations would be identity-preserving, too. Because they preserve an identity, they do not offend those with disability in the same way as prenatal testing.

If chronic, acquired conditions are ‘identity-restricting’, then is DS identity-restricting too? The limitations which DS confers make up an irremovable, life-long, significant influence on identity. The individual with a significant lifelong impairment cannot conceive of themselves without their impairment (Edwards, 2005). DS is partially and irreplaceably ‘identity-forming’, but is not identity-restricting.

Thus if numbers of those with DS fall, through parental choice or otherwise, this can logically be regarded as an affront to those with DS, as women are choosing not to have ‘children [somewhat] like them’. But how large is this affront? And are there other ways in which this hurt could be received?

4.3.3 Down syndrome and the use of a cure

For many years there has been research into the specific causes of DS. From this it is hoped will arise therapeutic interventions, and ultimately a cure (Dierssen, 2012). But does our search for a cure not offend those with DS in a similar way to active prenatal prevention? And would the use of a cure not replace individuals with
fundamentally different identities? Consider the following scenario, detailed by Gavaghan (2010):

Ursula and Aldous the parents of a newborn child, Isaac, are informed that he has trisomy 21, better known as [DS]. They are initially shocked, but come to accept and love Isaac, frequently marvelling at his capacity to surprise them. Four years later, Ursula and Aldous are informed by their GP that a new form of ‘gene therapy’ has been developed that can substantially ‘remedy’ the DS. If this is given to Isaac as soon as possible, he will develop ‘normally’. (pp.430-431)

The scenario Gavaghan (2010) presents is intriguing, and not entirely hypothetical- as recent research to treat DS shows some promise (Dierssen, 2012). How, then, should the parents proceed? Would we believe that they are acting negligently if they chose to withhold the treatment?

Negligence could be proposed, only if it could be proven that the parents are preventing the child from living a better life. But as discussed in Chapter 3.1, we do not have any workable definition for the good life. Why, then, do we not allow parents to prevent the setting of a broken limb- resulting in physical disability (Gavaghan, 2010)? It is hard to assert that a life with a malformed leg is necessarily worse than that of a person with functioning legs. Here the distinction I made in the previous section is important. The acquired loss of function resulting from not setting a broken limb is undesirable as it constrains an already-cemented sense of self. Setting a broken limb is an identity-preserving intervention.

But still the question remains, would it be permissible for the parents to refuse treatment? It is possible that Ursula and Aldous would declare that they love Isaac as he is, and have no wish to exchange him for another with greater intellectual ability. Is this a plausible defence?

Such a statement proposes that this ‘gene therapy’ would replace Isaac with another ‘self’. This not only agrees with the idea that DS is ‘identity-forming’, but asserts that DS is ‘identity-defining’, an integral part of who Isaac is such that if it is removed, Isaac will remain only in name. As previously discussed, this approach is problematic. It suggests that all people with DS are consistently similar, a statement which is incorrect.
This so called ‘replacement’ approach conforms to a largely cognitively-dependent view of personal identity (Gavaghan, 2010). It neglects the importance of narrative on personal identity espoused by John Locke, the relevance of an overlapping chain of distinct memories valued by Parfit (1984), and the significance of social interaction proposed by Gillett (2008). There is, of course, a possibility that memory is erased by the ‘gene therapy’, or it is left complete, or some memories take precedence over others. If memory is altered, however, is this important in a four-year old? Is it reasonable to suggest that at this age that self-concept is still in a very early stage of development and a change to part of this would be acceptable, in a way that the treatment for a 30 year old would not be?

Assuming a significant disruption to memory narrative, is this an identity-altering intervention? How does this treatment compare to the identity-restoring treatments aimed at curing Alzheimer’s disease? It seems that due to the great change in cognitive ability, with or without loss in memory narrative, this treatment would cause a large alteration in the identity-forming qualities of DS, and thus a significant change to the individual's self-identity, too.

If one’s self-identity was altered significantly by the treatment, could it be legitimately withheld by parents? Indeed, would it be wrong to administer the treatment? I would argue that given that DS is identity-forming but does not define the individual then it is reasonable to conclude that the change in personal identity will not be sufficient to significantly ‘replace’ a child like Isaac with another. However, the child would still be different. Because living with DS does not constitute a harm, and because DS is so varied in its severity, it is inappropriate for others such as the state to decide what DS means for a child’s personal identity. Parents are in the best position to choose whether the limitations conferred by DS are undesirable, or merely another challenge to be met.

Through this thinking, parents would neither be forced to use nor barred from using treatment; it would be their own choice. This conclusion is analogous to whether or not it is morally permissible to abort a fetus with DS; the decision must be made by the parents themselves. It is of interest that a similar argument supports parents’ choice to administer or withhold cochlear implants (Sparrow, 2005).
4.3.4 Assessing the link between expressivism and identity

Using a novel and mixed approach to identity, I suggest that DS will form part of one's personal identity in every circumstance. DS is more identity-forming than many other conditions, as it is present throughout life and presents with significant cognitive impairment. Because of this, a hypothetical ‘miracle cure’ for DS would be an identity-altering intervention, and not necessarily in the best interests of the child.

If DS is identity-forming, does this mean the expressivist argument when directed at prevention is valid? Partially, yes. But because DS is identity-forming and not identity-defining, the expressivist message is weakened. Furthermore, prevention of lives with DS by selective abortion is not the only way where numbers of people with DS could fall. I propose that the use of a cure could send similar ‘messages’, giving people with DS and others similar grounds for complaint. In fact, one could argue that the rejection of DS is more emphatic following treatment, especially if a cure was chosen by adults with DS themselves. This is in contrast to selective abortion, which is the result of a number of influencing factors for parents and not necessarily a rejection of DS itself (as discussed in Chapters 3.2.1 and 4.1.1).

Considering that treatment for DS may be more hurtful than prenatal testing, how far does this complaint extend? We would be unlikely to prevent those with DS the therapeutic choice of reducing the severity of their condition for fear of sending messages which ‘hurt’ others. In the same way we should hesitate to constrain reproductive choice through the banning of prenatal testing for DS.

Given that DS is identity-forming in a way similar to other disabilities (such as deafness, or spina bifida) does this mean that all with DS form a recognizable social group? Some anti-testing lobby groups oppose prenatal testing as they believe it specifically targets those with DS (Saving Downs, 2012b). But are those with DS necessarily a cohesive whole?

The anti-testing group ‘Saving Downs’ (2012b) believes that those with DS satisfy the criterion of a group due to their shared extra 21st chromosome, and their shared physical characteristics. This amounts to saying that ‘those with DS are more alike than they are different’. It rejects the notion that some with DS may not feel, or may not want to be, part of such a group. The grouping approach runs counterpoint to many disability
rights movements which stress that disabled persons are not their disabilities (Support Advanced Learning and Training Opportunities (SALTO), 2006). Organisations such as the New Zealand Down Syndrome Association (n.d.) emphasise that those with DS are individuals, and look and act more like their family members than others with DS.

Though those with DS may share some similarities, labelling all with DS as part of a distinct social group seems comparable to saying that ‘all those with glasses are part of a social group’. It is an unhelpful approach which prevents us from appreciating the diversity of all people in life, by passing judgement on a person because of the expression of a few traits. Because of this, those with DS should not be considered as forming a discrete social group.

### 4.4 An evaluation of the expressivist argument

Because DS can be considered as identity-forming, a health programme which actively aims to decrease the numbers of people with DS can be perceived as devaluing to those with DS, and those who represent the DS community. Thus, the expressivist argument when aimed at testing which specifically aims to reduce numbers of those with DS is at least partially valid. Therefore the aim of most prenatal testing programmes should not be to reduce DS birth prevalence but to facilitate women’s choice in an effective way. In short, public screening policies must not be construed as preventative.

This is true of screening in New Zealand and elsewhere, where prevention is not the aim of the programme (Antenatal Down Syndrome Advisory Group, 2007). However, the process of testing and selective abortion may still express discriminatory attitudes (Parens & Asch, 2000). Whether or not these hurtful attitudes are shared by those who offer or those who use the technology, a hurtful message is still received, as Holm (2008) has observed:

> Those who witness an action or utterance will often make inferences concerning what the action is meant to express. These inferences may be wrong, but as long as they are epistemically warranted, the witnesses are justified in claiming that the action did express this or that. (p. 25)
The perception that prenatal testing sends a message that ‘people do not value those with DS’ is partially epistemically warranted, due to the identity-forming nature of DS. Again, this does not mean that the inferences that are taken from the use of prenatal testing are correct, for testing and abortion can be for any number of reasons (as discussed in Chapter 4.1.1). But it does suggest that a message can logically be received. However, I believe the need to prevent this message by restricting prenatal testing is overstated, when weighed against other moral considerations.

4.4.1 The weight of hurtful messages

Timothy Murphy (2011) concludes that the weight of hurtful messages is not sufficient to restrict parents’ choices on testing and abortion. This is because the ‘hurt’ is indirect, and cannot be quantified. Thus, Murphy asserts, the consequentialist version of the expressivist argument is unproven, and should not influence policy.

The hurt is indirect for several reasons. Firstly, reproductive choices are not made by parents in direct view of those with disabilities; the termination of a pregnancy is a private matter for the family involved. Second, testing does not necessarily mean a prejudiced view of those with disability; a family with one child with DS may choose testing to ensure a non-DS child, and still love all resulting children equally. Thirdly, testing does not directly interfere with the ability of those with DS to engage in family and societal life, receive education and healthcare or pursue work opportunities. That is, the hurt does not translate into an actual direct ‘harm’ (Murphy, 2011).

Nelson (2007) agrees with Murphy, believing that for the consequentialist expressivist argument to be valid, one must prove that “had there been no social practice of prenatal testing and termination, people with disabilities would, other things being equal, have experienced less stigmatization.” (p. 476). Judging from the last 50 years, and the rise of disability advocacy, this is unlikely to be the case. However, it is still possible to assert that prenatal testing has functioned as a brake on the acceptance of those with DS and other disabilities, and may forever preclude their full acceptance. Gavaghan (2007) believes that this is unavoidable, and that if prenatal testing were prohibited, such discriminatory attitudes would be expressed in other, potentially more damaging ways:
We must ask whether we offer much solace by denying access to the means of implementing or demonstrating that judgement [i.e. prenatal testing]. Presumably some disabled persons will still be aware, or at least highly suspicious, that such attitudes exist and that the only reason they are not routinely implemented is that 'society' in its other conception - the legislature, the courts and the regulatory bodies - prohibit them from being so. (Gavaghan, 2007 p. 28)

For Gavaghan (2007), these judgements will be evident in other ways - such as through lobbying, court action, public media, or travelling to other countries where prenatal testing remains legal. The expressivist argument will thus remain valid, but will be more direct and hurtful, due to the magnitude of the actions which could take place. Hence, preventing people from acting is not the way to change judgements and viewpoints, and it is these judgements which should be addressed directly.

Preventing one possible outlet for a discriminatory judgement does not prevent it from being expressed in other ways. With this in mind, the expressivist argument is more an argument against discriminatory attitudes than against the practice of testing and abortion per se. The best way to address the expressivist argument is to address these attitudes, not to deny women access to prenatal testing and selective abortion.

4.5 Conclusion

The expressivist argument is helpful as it encourages us to examine the structures, processes and choices which surround prenatal testing and selective abortion. It persuades us to think twice about our actions and the meanings and rationale behind them.

I accept that the prevention of DS can be seen as hurtful to people with DS, as it prevents the birth of 'more people like them'. For this reason, public health policies should not aim to prevent births with DS. DS is only partially identity-forming, and thus the expressivist argument is weaker than if people with disabilities 'were their disabilities'. It is not so weak, though, to claim that prenatal testing offends those with DS in the same way that abortion offends those who are 'pro-life'.
However, the consequentialist version of the expressivist argument is unconvincing. The quality of lives of those with DS and disability and the use of prenatal testing have both increased in unison. Though testing for DS may be a further barrier to inclusion, little evidence for this exists.

The non-consequentialist version of the expressivist argument is more robust, as many groups do feel hurt by the practice of testing and abortion. However, it is doubtful that people should feel hurt; as state policies are aimed at maximising individual choice, and an individual’s choice is a complex decision made up of many factors. But regardless of whether people should feel hurt, people do. Although this hurt exists, it can be mitigated through other paths, namely via vocal recognition and continuing support of the DS and disabled community.

The expressivist argument can apply to all forms of genetic selection. Indeed, it can apply to many actions in pregnancy, as these are also actions taken to avoid the raising of a disabled child. However, it is pertinent to point out that these and other behaviours are supported by bodies which represent such groups. One example is the New Zealand Organisation for Rare Disorders, which strongly advocates for the mandatory fortification of bread with folic acid to prevent the incidence of neural tube defects (Levy, 2012).

The use of a cure could also express similar judgements, which could be all the more hurtful if chosen by people with DS themselves. Many organizations which represent disabilities actively support research for a cure. The US-based ‘National Tay-Sachs & Allied Diseases Association’ provides support to parents of children with Tay-Sachs and other diseases, “leading the fight to treat and cure Tay-Sachs, Canavan and related diseases” (National Tay-Sachs & Allied Diseases, n.d.).

Some organizations such as Down’s Syndrome Scotland believe that prenatal testing should remain a choice for parents (Summerfield, 2009). Others oppose prenatal testing and abortion, yet support further research for treatment and cure. As both research and testing are in some way expressivist, I suggest that supporting one and not the other is inconsistent. Organizations can both care for those with DS, and appreciate the role of testing; these two views are not contradictory.
Part Three: Questions of policy
Chapter 5 \hspace{1em} THE SYSTEM AND THE PROCESS: TESTING AS IT IS, AND TESTING AS IT SHOULD BE

I have suggested that selective abortion for DS is morally permissible and the offer of testing and abortion morally justified. But this does not mean that present methods of testing are ethically sound. This chapter examines areas of testing which some have suggested are problematic. These areas include the way in which testing is presented to women, influenced in part by health professionals and their views towards disability. Other issues include the routinisation of prenatal testing for DS, and the possible drop in numbers of people with DS we may see in the future. Finally, I propose a method with which to evaluate other forms of prenatal testing.

5.1 The views of health professionals

Some objections to prenatal testing are aimed at the way in which testing is presented to pregnant women. If DS is presented as undesirable, and abortion medically necessary, one would expect a woman faced with a positive result to abort regardless of her wishes. Some have argued that there exists ‘a medical culture that is passive if not indifferent to disability issues’ (Klein, 2011 p. 15). If the doctors, nurses, midwives and genetic counsellors hold negative views towards the conditions being tested then, it is theorized, they will act persuasively- whether actively or passively- to try and prevent the woman from continuing pregnancy.

Because levels of indecision about the options opened up by prenatal testing are frequently high (Dery et al., 2008), the way that abortion and disability is framed by health professionals may have important repercussions for women's attitudes to disability and pregnancy (Klein, 2011). What is the evidence that discriminatory attitudes permeate the thinking of health professionals, existing in the systems and processes which surround prenatal testing?

It is true that clinicians may have different attitudes towards prenatal testing and abortion than patients. Studies in Israel (Dery et al., 2008) and Europe (Drake et al., 1996) have shown that maternal care providers (including paediatricians, geneticists, obstetricians and nurses) are more likely than members of the public to support pregnancy termination for all fetal abnormalities. Similar results were found by Dorothy
Wertz (2000) when data were collected on the attitudes of physicians, genetics professionals and mothers of children. Wertz found that 62% of physicians would abort in the case of DS, compared with only 36% of the mothers with children. Genetics professionals were the most likely to favour termination of pregnancy, with 80% supporting abortion. This pattern was repeated in almost all of the 22 other diseases assessed by Wertz, with geneticists and physicians always favouring abortion significantly more than mothers.

Wertz’s (2000) study was not without its limitations. The comparison group of ‘mothers of children’ is not necessarily reflective of views of the public due to various factors such as income, age and gender. Furthermore, the study was localised to the US and may not reflect the views of the public (or health professionals) in other Western countries. However, the significant difference in attitudes corroborates earlier evidence that healthcare providers favour testing and termination more than members of the public (Wertz, 1998).

Are these views necessarily ill-founded and discriminatory? Medical practitioners and geneticists have more interaction with people with disabilities than those of the general population. However, these health professionals often see the negative side of disability - that which requires frequent visits to the hospital, extra care and support, and significant limitations. It is hard to conclude that health practitioners’ views are discriminatory, if they see the reality of living with impairment every day. It is more likely that some medical professionals overestimate the burden of disability, but this does not mean this is discrimination.

However, placing undue emphasis on the burden of disability is still problematic, especially if these views are conveyed to patients faced with the choice of prenatal testing. If this is the case, one would expect more women to choose and act on testing. Van der Berg et al. (2007) investigated whether midwives’ and physicians’ attitudes towards testing influence pregnant women’s decisions and attitudes towards screening. Their findings were based on a large sample size of 945 pregnant women and 97 health professionals. Results suggested that women’s choices to use testing, or their attitudes towards testing, were not predicted by their health professional’s views. This suggests that although maternity carers may hold different attitudes to testing and abortion, this does not- actively or passively- persuade women to engage in prenatal testing. However,
this study occurred before the advent of routine prenatal testing, and reminded both parties that testing was the women’s own choice.

A study of women who continued their pregnancy after a diagnosis of DS indicated that the majority had experienced what they perceived as encouragement to have amniocentesis (Skotko, 2005). Significantly, a small number even felt pressured (Skotko, 2005). These findings are corroborated by Wertz’s study (2000) which found that 13% of physicians emphasized the negative aspects of DS to encourage women to terminate, without suggesting it directly. However, the reverse was also true; 10% of physicians emphasized the positive aspects of DS, encouraging women to continue pregnancy. Of particular significance was the finding that 10% of respondents actively urged termination.

These studies do not suggest that persuasion of women to test and abort in the case of prenatally diagnosable conditions is the norm. This is encouraging, indicating that the majority of women are facilitated in their decision-making. However, Wertz’s (2000) findings suggest that a small yet significant number of physicians allow their own preferences and views to interfere with their responsibility to provide objective information. Non-directive counselling is the ideal, and when faced with a positive result both persuasion to keep or persuasion to end the pregnancy is equally undesirable.

5.1.1 Attitudes of the medical workforce towards impairment: Inadequate, or imperfect?

It is clear that physicians, midwives, nurses and medical geneticists have views on impairment that are different to that of the general public. This does not mean that their views are ill-founded; in fact, some may have well-informed views of what a disability like DS means for both family and individual.

In a more positive light, most pregnant women are happy with the care they receive from health staff. A study in Sweden showed that 88% of those who terminated in the case of DS were ‘considerably satisfied’ with their contact with nursing staff or obstetricians, and 71% were considerably satisfied with midwives (Korenromp et al., 2007).

It seems clear that a small number of women are not well cared for under current approaches to prenatal testing. However, for most women, the availability and use of
testing is a positive experience. Thus, the service at present may not be optimal but it is adequate and addresses most people’s needs. This does not mean that efforts should be directed away from accomplishing best practice. But it does mean that objections based on the idea of the health workforce as pervasively stigmatizing and discriminating should be rejected.

An area in need of improvement is medical education. Some studies have shown that students of health professions hold negative attitudes towards those with disabilities (Tervo & Palmer, 2004). A survey of 532 obstetricians and gynaecologists in the US found that although over 80% felt that they had the necessary skills to counsel patients on genetic issues, 45% of the physicians rated their postgraduate training on prenatal diagnosis as barely adequate or non-existent (Cleary-Goldman et al., 2006). That medical practitioners feel that their training is so lacking is problematic, leading to poor outcomes for both doctors and patients.

Some progress has been made. Generally there has been improvement in physician attitudes and behaviours in the last few decades (Klein, 2011). Recently there has been increased recognition of the lack of education for medical students and practitioners (Skotko, 2011). More comprehensive training has been developed, such as online simulations for physicians to practise prenatal testing and their options (Ferguson et al., 2006), as well as opportunities for medical students to interact with people who have DS (Pfeiffer, 2009). In some medical schools, there have been significant changes to medical curricula to make them more disability-minded (Symons et al., 2009). These interventions are positive, not only for the non-directive offer of prenatal testing services, but also for the general management and interaction with people with disabilities in general.

Though there are undoubtedly some problems with the services and processes surrounding prenatal testing, these are problems to be fixed and do not warrant further restriction of testing. However, some such as Nancy Press (2000) and Sonia Suter (2002) assert that it is not only health professionals and their views that are problematic, but that the positing of prenatal testing in routine medical practice compels women to take part. What is the argument which supports this, and is such ‘routinisation’ necessarily a bad thing?
5.2 Routinisation and Medicalisation

Pregnancy is, by all accounts, a perfectly natural process. Yet it is a process which has progressively come under the close scrutiny of the medical profession. Pregnancy is increasingly seen as a ‘risky’ process, one which often requires a large number of interventions. Such interventions might include dietary changes for the mother (avoidance of certain foods, reduced intake of alcohol, supplementation of vitamins), avoidance of some activities (such as spa pools or saunas) and frequent check-ups with health professionals (general practitioners, midwives and obstetricians).

The frequency of pregnancy-related interventions has increased over time; for example, the incidence of Caesarean section has been increasing worldwide for many years (Burrow, 2012). Some argue that the increasing use of such interventions creates a ‘normalisation’ of the technology, forcing patients to expect more and pressuring medical staff to fulfil ever-increasing expectations. Some feel that women are influenced by ‘the technological imperative’; the compulsion to use technology merely because it is available (Sherwin, 2000).

Prenatal screening for DS and other conditions is particularly at risk of becoming routine, for it is sometimes offered as one test amongst a dizzying barrage of pregnancy related procedures. Many similar-looking tests are routinely offered during pregnancy; such as full blood counts and typing, and testing for rubella, hepatitis B, syphilis and HIV (Up To Date, 2012b). The material required for these tests is exactly the same; just a small amount of blood. Yet the implications of the tests are very different.

Routine antenatal screening for conditions like rubella functions to ensure the health of the fetus, and enables diagnosis and treatment for the mother. It is therefore preventative. Prenatal testing for DS and other conditions is not. Though screening tests may appear very similar to other antenatal procedures they enable choices which ultimately decide the course of pregnancy, not ensure pregnancy continuation.

Ultrasound is another example of a test that has become routinely accepted, though the wider implications of the test are sometimes not well understood. Ultrasound is commonly used to tell the sex of the fetus. For some women, the first test is looked forward to with expectation and excitement, marking an emotional signpost of pregnancy because of its power to visualise the fetus (Seavilleklein, 2009). However, the
medical use of ultrasound is less benign; it functions to detect developmental abnormality in the fetus, and is frequently used to increase the accuracy of DS screening. Ultrasound is currently a routine part of pregnancy management, commonly offered at 19 weeks, sometimes with a previous offer around week 11. In some countries such as the UK, women have typically received two or three scans by the end of pregnancy (Whynes, 2002).

Like ultrasound, prenatal screening is particularly vulnerable to being seen as ‘just another test’. This process has already been observed in the use of the maternal serum alpha fetoprotein (MSAFP) testing. In the past, measuring MSAFP was the most common way of screening for fetal anomaly. It is now used to supplement some forms of DS screening.

In general, screening is offered to all women regardless of age. MSAFP screening became a routine offer of testing, in the same way that offering DS screening is routine in some countries today. Some found the offer of MSAFP screening hard to refuse, feeling that it “encouraged [women] to begin down a path that can lead to a selective abortion without having to... think through, or face this endpoint” (Press, 2000, p. 221).

It is hard to argue that obscuring the details of the follow-up of any screening programme is justified. Fetal screening programmes should be transparent, as they ultimately result in ‘life or no life’ decisions. The routinisation of the MSAFP test is problematic, made so because little attempt was made to highlight the differences of this kind of test. In most countries with screening for DS and other conditions, pamphlets and other informational material along with a consult with a health professional are provided before testing. This is one way in which the link between screening and abortion is not clouded from view.

It is not possible to definitively conclude that the routinisation of prenatal testing has contributed to a marked increase in the uptake of testing. A trial at one UK hospital (Klein, 2011) suggested that there was no discernible difference in uptake rates of testing when DS screening was offered in conjunction with other routine blood tests. Though not refuting the routinisation argument, this does suggest that many women do distinguish between routine tests for pregnancy and prenatal testing for DS.
Nevertheless, problems remain. One study of 982 pregnant women found that only 43.5% of women made an informed choice regarding DS screening (Dormandy et al., 2006). A major reason was poor knowledge about DS screening itself. This suggests that at this hospital at least, rates for informed choice regarding DS screening are unacceptably low. It is of note that the risk of being uninformed was equal among those who chose to have testing and those who declined.

As the biggest factor influencing uninformed choice appears to be a lack of knowledge regarding DS screening, it is clear that more informational materials should be provided, and greater efforts made to make sure these materials are understood.

5.2.1 Evaluating the weight of ‘routinisation’ arguments

Some commentators feel that the combined factors of poor information, discriminatory attitudes, pressure by health professionals and the routinisation of prenatal testing summate to reduce women’s reproductive autonomy and increase the rates of selective abortion (Klein, 2011; Suter, 2002). Though I have found some evidence that these factors are present, the argument that current prenatal testing actively violates a woman’s autonomy (Klein, 2011; Seavilleklein, 2009) is unconvincing. This is because the mere availability of prenatal testing enables more control over one’s life and parental prospects. Purely because the process of informed consent is not perfect does not mean that a choice is predetermined by the health service. The system at present allows women a choice, though improvement in the way prenatal testing is offered could make this choice more open and informed.

Support for this argument is shown by uptake rates for DS screening in New Zealand. A screening programme for DS and other conditions was introduced here in 2010. Only 55% of pregnant women decided to have prenatal screening in 2011 despite it being offered to all pregnant women (National Screening Unit, 2012). This points to fair and unpressured choices by women. If reproductive autonomy is jeopardized by the current screening procedure, one might expect the number ‘conforming’ to screening to be much greater. This is not to say that all screening programmes are free of some degree of active or passive pressure. However, it suggests that even in a nationwide screening programme, the power of the ‘technological imperative’ is not as great as might be imagined.
Because an intervention is routine does not necessarily mean that it is problematic. The vast majority of medical interventions which comprise routine medical practice have helped maternal and fetal mortality to fall significantly. However, it is worrying if women feel forced to engage in prenatal testing just because it exists, or because they will be seen as irresponsible if they do not. As argued above, evidence indicates that most women do not feel this way, and with better information which emphasizes unequivocal acceptance of parental choice this issue should be minimised.

5.3 The end of Down syndrome?

The use of prenatal testing continues to increase, and it is possible that in the future there may be fewer individuals with DS than there are today. As outlined in Chapter 2.4, tests such as Non Invasive Prenatal Diagnosis (NIPD) may markedly increase the number of DS terminations. Eventually, we may reach a society where congenital disabilities like DS are very rare indeed.

As I have written elsewhere (Cole & Jones, 2013), it is unlikely that DS will disappear (see Appendix). Testing is unlikely to become mandatory for several reasons: because this would be an unjustified interference of the state into one’s own reproductive determination, because this would be startlingly similar to past regimes of state-controlled eugenics, and because most countries hold informed consent as crucial to the delivery of modern healthcare. Thus, there will always be women who decline testing for DS, or decline pregnancy termination. Furthermore, some abnormalities escape detection using even the most accurate diagnostic techniques.

Nevertheless, it is likely that at some time in the future, numbers of people with DS will drop. Should testing be restricted, to prevent this from happening? It is hard to justify that women have a responsibility to maintain the existence of DS in future generations. As Murphy (2011) has written, “there is no requirement that [those with disabilities] exist as either a fixed number in the population, any more than there is a moral requirement that there can be specific numbers of other kinds of people” (p. 107) such as those of a certain height, or people with exceptionally high intelligence, or believers in vanishing religions.

The distribution of human qualities is continually changing, and there is no reason that one should be held constant (other than, possibly, sex ratios). Because it is
hard to say that the exact composition of today’s society is any better than previous or future societies, it should not follow that women have a moral obligation to reproduce in a way to replicate this.

5.3.1 A reduction in numbers, and a loss of support?

Some contest that with a drop in numbers of people with DS there will be reduced social resources and consideration for such people, and this would constitute an actual harm (Callahan, 1995; Shakespeare, 1995). This is what has been termed the "loss of support" objection (Buchanan, 1996, p. 21). This may seem plausible, as it might follow that there would not be as many wheelchair access ramps, if there was a tenth the number of wheelchair users (Gavaghan, 2007). Furthermore, less research may be allocated to find a cure for only a few dozen people (Buchanan et al., 2000). As the case of the Pharmaceutical Management Agency (PHARMAC) in New Zealand shows, it is true that very rare disorders do not always receive optimum treatment (Newtonhealth, 2011).

However, it is also possible that fewer people with DS could mean that resources could be spread more evenly, with each individual gaining more. This has been the case with a reduction in thalassemia, a disease which affects the blood (Kitcher, 1996). People with DS could benefit with better treatment for co-morbidity, and more individualised support for education and family, with a reduction in the prevalence of DS.

It is possible that with less numbers of people with DS, network and support groups (such as the New Zealand Down Syndrome Association) could become weaker, and the community that one enters when raising a child with DS could become smaller and more localised. However, with the huge potential for information dissemination offered by the internet, a small group could still be very effective. Indeed, not all those who raise a child with DS wish to become a part of such groups. Irrespective of such considerations, it is untenable to suggest that pregnant women have an obligation to ensure that these groups continue to function.

Even if we concede that those with DS may be harmed in some way in the future via a reduction in their number, it does not mean that stopping or restricting prenatal testing is the answer. It would make far more sense to continue to monitor the support
for those with DS, and increase it if necessary. As Murphy (2011) has written, even if a direct link between prenatal testing and a harder life with DS or disability was proven, other avenues to better these lives should be pursued, rather than reflexively banning testing. Such measures could include anti-discrimination laws, or positive advertising.

The offer of testing should not mean that we cease to support affected individuals and their families. Historically, the converse is true; political consideration and respect for people with disabilities have been on the rise since the disability rights movement of the 1960s, at a similar time when amniocentesis was first introduced.

Treatment for all those with disabilities is almost definitely improving (Murphy, 2011). Seminal legislation such as the Americans with Disabilities Act 1990, followed by the United Nations Convention on Human Rights of Persons with Disabilities 2006, discourages discrimination and aims to ensure equality. These well-considered documents send a strong message that people with disabilities are a valued part of society. Indeed, some commentators label the disability rights movement as a fundamental part of the civil rights movement- a feat “that is arguably the greatest moral achievement of the twentieth century” (Parens & Asch, 2000, p. 37). This has been in tandem with the development of reproductive technologies, not opposed by it.

5.4 Different disability, different disease: Where to draw the line

For many years there has been considerable debate on the potential of the ‘new genetics’ to test for a multitude of diseases. Though amniocentesis has been practised since the 1960s, karyotyping remains limited to detecting large-scale chromosomal alterations, such as trisomy 13 and 18, and DS. Now, other methods of genetic testing allow a raft of disorders such as Huntington disease, Fragile X syndrome and cystic fibrosis to be tested for in the developing fetus. Many more tests are being developed.

What diseases should and should not be tested for? Or should there be no restrictions, with parents able to make their own decisions? Though there may be more pressing grounds for testing some diseases than others, no consistent framework has been proposed which has avoided objection. Jeffrey Botkin (1995) proposes four criteria when considering a condition for prenatal testing; the severity of the condition, the age of onset, the risk of the genetic predisposition being expressed as disease, and the probability that the disease will occur in people without a genetic predisposition. Botkin
asserts that the severity of the condition should be viewed as the severity to the parents; i.e., the greater demands for care required by the disorder. This is, ultimately, related to the severity of the disease.

From this, Botkin (1995) proposes four general categories of conditions which would fulfil these criteria. These are conditions which are fatal in childhood, those where a child will experience significant suffering and require hospitalization, conditions that will not permit independence in adulthood, and conditions where demands on the parents are very large. Following these criteria, testing for DS would be permitted as dependence in adulthood and greater parental demands are implicated in the condition. Examples of other diseases which meet Botkin’s criteria and where testing should be permitted are shown in Table 1.

<table>
<thead>
<tr>
<th>Factor VIII and IX deficiency</th>
<th>Sickle cell anaemia</th>
<th>Menkes syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fragile X syndrome</td>
<td>Cystic fibrosis</td>
<td>Tay-Sachs disease</td>
</tr>
<tr>
<td>Lesch-Nyhan syndrome</td>
<td>Spina bifida</td>
<td>Trisomies 13 and 18</td>
</tr>
<tr>
<td>Hurler syndrome</td>
<td>Other inborn errors of metabolism</td>
<td>Major structural abnormality</td>
</tr>
</tbody>
</table>

Table 1 Diseases suitable for testing under Botkin’s criteria. Based on (Botkin, 1995)

Many of these conditions are currently tested for in many parts of the world. Because some of the conditions have a low incidence population-wide screening is not usually undertaken, except for the notable exception of Tay-Sachs screening amongst Ashkenazi Jews. Instead, testing is offered if there is a family history of the disease.

In light of the arguments I have adduced, the diseases included here are morally test-worthy (but I assert that this is not necessarily a comprehensive list). Many of these conditions differ from DS in that they have a markedly reduced lifespan (Lesch-Nyhan, Tay-Sachs and cystic fibrosis are all examples). Many involve significant, repeated episodes of pain and suffering (spina bifida, sickle cell anaemia, Hurler syndrome). Both of these qualities are consistent with a restriction of an open future and living a less good life, and in principle can be used to justify abortion. DS does not have a markedly reduced lifespan (life expectancy is around 60) or significant pain. The conditions shown in Table 1 constitute at least as good reasons for abortion and testing as DS (with the possible exception of Factor VIII and Factor IX deficiency).
What, then, would be examples of medical conditions where abortion would not be justified? Botkin (1995) asserts that there are five classes of conditions where testing should not be offered; those that are probably treatable given early intervention (phenylketonuria, asthma, cleft lip and palate), conditions which experience limited suffering and hospitalization (many thalassemias, Marfan syndrome, achondroplasia), late onset conditions (polycystic kidney disease and Huntington disease), genetic predispositions which involve a significant proportion of risk (ie genes for schizophrenia, depression and obesity) and natural forms of human variation (low height, eye colour, sex of the fetus).

A criticism of Botkin’s (1995) approach is that it attempts to make concrete distinctions between ‘serious’ (such as DS and Tay-Sachs) and ‘non-serious’ diseases (phenylketonuria, achondroplasia). By defining some conditions as worth testing for and some as benign, those in the serious category may feel that part of who they are is seriously worth avoiding (i.e., the expressivist argument applies) (Gavaghan, 2007). Vice versa, those with diseases labelled as ‘non-serious’ may feel that the significance of their disease is undermined. Some commentators argue that this will spread discord within the disability community (Parens & Asch, 2000).

Botkin (1995) argues that testing for natural forms of human variation should not be permitted. Some argue that all forms of disability are examples of normal human variation (Asch & Barlevy, 2012). In applying this stance to Botkin’s criteria, all forms of testing should be prohibited. I reject the all-encompassing human variation model, as most of the conditions in Table 1 are rare, involving a marked departure from normal structure or function, and are often associated with repeated episodes of pain and suffering. Consequently, they do not constitute typical forms of variation. Moreover, just because something is part of natural ‘human variation’ does not mean it is valuable; for example, most cancers and infections are naturally occurring, yet we seek to cure them. Conditions like Huntington Disease may be ‘natural’ but we still regard disease progression as full of suffering and pain. We can value and respect all forms of human difference, and still maintain that some conditions are undesirable.

The most important drawback with Botkin’s (1995) approach is that it neglects an individual woman’s situation; for example, it does not allow abortion for a fetus diagnosed with phenylketonuria even if the family are poor and unlikely to be able to
afford the high-cost and rigid diet that the child will require (Wertz, 2000). Furthermore, Botkin’s categorical approach to testing is at odds with the role of medical practitioners, who ideally should employ some measure of holistic evaluation to prenatal testing. This would take the subjective experience of the pregnant mother into account including relevant factors, such as family history and the ability to cope with the extra responsibility of care. Nevertheless, Botkin’s categories are important, and mirror the framework of prenatal testing in many countries today.

An alternative perspective is provided by Dorothy Wertz (2000), who offers a ‘harm/benefit’ approach to prenatal testing. She opposes the separation of diseases into serious and non-serious categories. Rather, she differentiates women into those that are ‘high risk’ (if one has a family history or advanced maternal age) and should be tested, and those who are ‘low risk’ (all other women) and should not be offered testing. Other tests may be carried out routinely, if the identification of the disorder will be beneficial for the fetus (e.g. by allowing early intervention). If a woman is not ‘high risk’, then she must prove that significant harm (i.e. psychological distress) will occur if testing is not carried out.

Wertz's (2000) criteria are helpful as they avoid the definition of what is a serious condition, and allow a more tailored approach to which particular tests are offered to each pregnant woman. However, by avoiding setting limits on what can and cannot be tested for, the system is open to abuse. If one has a family history of asthma, should one be able to test and then terminate on the presence of asthma in the fetus? This gives the distinct impression of being a frivolous, irresponsible use of such a technology and should not be employed as a reason for abortion.

Furthermore, Wertz’s (2000) criteria do not permit tests for Botkin’s (1995) serious conditions for the majority of women, as the majority of women are not ‘high risk’. Wertz would therefore reject screening for DS and other conditions, though they are morally justified. Wertz’s ideal of individualised testing is flawed, as many women do not know what they are at risk of, may not be aware of their family history, or recognise what tests are available to them. In short, not all women know what tests are best for them.

A libertarian approach rejects any constraint on what prenatal tests should be available to women, asserting that the separation into ‘serious’ and ‘non-serious’
categories is untenable. Women, it is argued, should have their own input into what constitutes as ‘serious’ for them. This is inevitably subjective and related to culture, socioeconomic status, religion, and other values (Gavaghan, 2007).

This position accepts that in the final analysis, women alone will decide whether the condition has the capacity to significantly impact on the child’s chance to lead a good life. Thus, women should be able to test for whatever conditions they desire; from serious congenital conditions like Tay Sachs, to an increased likelihood of developing dementia with age, or schizophrenia in early adulthood. This is epitomized by Savulescu (1999), who goes as far as to permit sex selection for family balancing, suggesting that early abortions could be permitted on the basis of fetal sex alone.

By allowing unfettered access to testing one allows the option of abortion for insubstantial reasons. If we regard the fetus as having an increasing right to life, then the reason for its termination must be greater than its current right to life. It is misdirected to terminate a pregnancy on the grounds of, say, a future child’s increased risk of developing Alzheimer’s. By using these tests one treats the natural process of pregnancy like a fact-finding mission. The fetus’ genetic privacy is breached, as in the case of continuation of pregnancy, the parents will know of predispositions that the child may not wish others (or themselves) to know. By permitting tests which are allowing a preference instead of fulfilling an understandable requirement the value of the process of pregnancy and respect for the developing fetus is lost.

With this in mind, it is unlikely that many would test a fetus for its predisposition to asthma, say. Nevertheless it could occur, which is both a pointless exercise and a flippant use of technology. Because some tests should not be permitted, limits on prenatal testing must be defined.

5.4.1 Theory from a thesis: How an examination of testing for Down syndrome can inform wider policy

In general, I find myself in agreement with Botkin’s (1995) disorders deemed suitable for prenatal testing. However, I believe that Botkin is too fixated on the medical aspect of disease. An approach is required which focuses on the experience of living with the disorder- for both child and family. I have previously made the point that the
decision to abort should not be purely medical, but should be holistic, encompassing many values. Thus, broader criteria for prenatal testing are warranted.

As mentioned in Chapter 4, the justification for prenatal testing is dependent on the permissibility of selective abortion for the disorder in question. Thus, I have developed a brief set of criteria which reflect this. Those relevant to the moral permissibility of abortion are covered in criterion A. Some forms of test do not enable abortion, but are of direct benefit to mother or fetus. These tests are sufficient under criterion B.

For a condition x, the criteria I propose are as follows:

A 1) Can condition x result in significant disruption to parents’ lives?

A 2) Can condition x significantly limit an open future?

A1 includes recognition of the extra demands made on parents to care for a child with x. A2 recognises the impairment, life expectancy and pain or suffering associated with condition x.

If both A1 and A2 are fulfilled then selective abortion for condition x is permissible. If selective abortion is permissible then testing to allow parents this option is fair. It does not necessarily follow that testing should be actively offered to all pregnant women; suffice to say that they have the right to access the test if requested.

If either A1 or A2 is not valid then selective abortion for condition x is impermissible. However there are some cases where the information from a prenatal test may be beneficial; by allowing medical interventions which may benefit the health of the fetus or mother, or psychological preparation for the birth of a child with significant illness. In these cases we turn to B:

B) Does the information provided by prenatal testing for x provide any significant benefit for the mother or fetus in any way?

If B is fulfilled but neither A1 nor A2 is valid then testing is justified, but any result is not a sufficient ground for abortion. The result is beneficial in improving outcomes for mother or fetus, and therefore testing can be permitted.
This is a pluralistic set of criteria for prenatal testing. It does not require a direct analysis of the medical ‘severity’ of the condition, instead using the way in which a condition affects lives (of the parents and of the fetus) to inform our judgement on testing. It should be noted that if effective treatment is available for condition x then a child’s ‘open future’ would be considered to be less limited, and condition x less suitable for prenatal testing. Some disorders and traits with the way in which they would be evaluated using the criteria are shown in Table 2.

<table>
<thead>
<tr>
<th>Condition</th>
<th>A 1) Can condition x result in significant disruption to parents’ lives?</th>
<th>A 2) Can condition x significantly limit an open future?</th>
<th>B) Does the information provided by prenatal testing provide any benefit for the mother or fetus?</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Down syndrome</strong></td>
<td>Yes, due to the increased parental demands for care, and relative dependency throughout life.</td>
<td>Yes, mainly due to the significant degree of cognitive impairment associated with DS. The many co-morbidities associated with the disease are also of note.</td>
<td>N/A</td>
</tr>
<tr>
<td><strong>Cystic fibrosis (a chronic lung disease, which also affects many parts of the body)</strong></td>
<td>Yes, due to increased parental demands for care and repeated hospitalizations, and some lack of independence.</td>
<td>Yes, due to reduced life expectancy and repeated hospitalizations.</td>
<td>N/A</td>
</tr>
<tr>
<td><strong>Achondroplasia (a disorder of bone growth and the most common type of dwarfism)</strong></td>
<td>No. Achondroplasia does not usually result in significant and frequent hospitalization or significant</td>
<td>No. Though those with achondroplasia may not have lives as ‘open’ as others, this limitation is not significant. The</td>
<td>No. This information is of no influence to prenatal care or delivery. It is doubtful that psychological preparation for birth</td>
</tr>
<tr>
<td>Disorder</td>
<td>Description</td>
<td>Impact on Parents</td>
<td></td>
</tr>
<tr>
<td>---------------------------</td>
<td>-----------------------------------------------------------------------------</td>
<td>-----------------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td><strong>Asthma</strong></td>
<td>No. Asthma is usually a mild disease which does not require significantly extra care by parents.</td>
<td>No. Those with asthma still have an open future.</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>No. This information is of no influence to prenatal care or delivery.</td>
<td></td>
</tr>
<tr>
<td><strong>Multiple pregnancy</strong></td>
<td>Yes. In some cases, multiple pregnancy may be hazardous to the mother’s health. Furthermore, the raising of a multiple pregnancy can significantly disrupt parents’ lives. Parental care will not be as lifelong as caring for a child with DS, however.</td>
<td>In some cases. Multiple pregnancies, especially those of higher orders, often result in premature birth, and have an increased risk of cognitive and physical impairment. The degree of risk will vary greatly, but in some cases will be highly likely to result in a condition which limits a child’s open future.</td>
<td>Yes. Identifying a multiple pregnancy may benefit both mother and children with more rigorous prenatal care and specialist supervision in delivery. Detection of multiple pregnancy may be helpful in preparing for increased parental demands of raising several children. The emotional link of ultrasound may also be of benefit.</td>
</tr>
<tr>
<td><strong>Female sex</strong></td>
<td>No. The raising of a girl confers no significant disruption to parents’ lives (in any culture).</td>
<td>No. Even in some countries where there is evident gender inequality (such as parts of India or China), females are not significantly limited in their future.</td>
<td>Yes. Ultrasound may provide a significant emotional link between mother and fetus. This test will often inform parents of the fetus’ sex.</td>
</tr>
</tbody>
</table>

Table 2 Various disorders and traits assessed through a framework for evaluating the suitability of prenatal testing
The criteria are relatively strict. They limit selective abortions to conditions which confer some measure of harm via a significant disruption to parents, and which make a significant disruption to an open future. It is suggested that most congenital conditions which result in significant cognitive impairment would be sufficient reason for abortion under this framework. It is conceded that in some cases of multiple pregnancy, especially those of higher orders, the magnitude of risk to mother and fetuses would be such that selective abortion would be permissible. Testing for multiple pregnancy has the added advantage of improved pre and peri-natal care, benefiting the health of mother and children.

Abortions would not be permitted for conditions which did not fulfil these criteria, such as achondroplasia or asthma, or traits such as female sex. However, testing could be justified if it was of benefit to fetus or mother. As shown in Table 2, this is the case for prenatal sex determination via ultrasound.

The above framework involves terms which are not explicitly defined, such as ‘significant’, and ‘open future’. For most disorders, the judgement will be relatively straightforward. For others, the judgement will be less clear. However, these criteria help to guide discussion to the areas which are most important when considering conditions for prenatal testing.

The most controversial criterion may be A 2, ‘can condition x significantly limit an open future?’ This criterion is founded on the notion that some life options are important and irreplaceable. Some will argue that having some life options is not important. Others will propose that we are all limited in our life options throughout life, from the point of conception. Though this is true, some are limited in more ways than others. Social circumstances, such as poverty, poor housing and unhappy upbringing limit life options in lesser ways than many of the conditions above. We oppose these circumstances, and act to ameliorate them through social welfare and other actions. Because we hold high the tenets of equality and justice, it is permissible to oppose the limitation of options from a condition of significant impairment. The impairment must be significant, however, as abortion is a value-negative action that requires strong grounds for its justification.

It should be reiterated that these criteria tell us which traits it is permitted to test for. How women react to a positive result is entirely their decision.
5.5 Conclusion

Though testing and selective abortion for DS is morally defensible, this is not true of some of the processes which surround prenatal testing. In many countries the system is not fundamentally flawed, yet it is not as good as it should be. Some health practitioners still hold negative views on people with disabilities. Inaccurate and unhelpful information about what DS means for a life sometimes surrounds DS testing. The choice to have testing and the choice to have an abortion is not a medical choice but a holistic one; one which ultimately involves the woman’s subjective conception of a good life. Information should reflect the experience of what it means to have DS, and not merely the increase in morbidity and mortality associated with the condition. Encouragingly, steps are being made in this direction.

Considerable controversy surrounds the advent of new prenatal tests. Some worry that soon parents will be testing for an increased risk of schizophrenia, or the presence of blue eyes, or a significant chance of homosexuality (Close Up, 2012). Testing for such conditions not only undermines the future child’s genetic privacy but also allows selective abortion for trivial conditions, under the guise of ‘social abortion’. By considering whether a condition significantly disrupts parents’ lives, and whether it significantly limits an open future, one will prohibit many forms of these tests. Botkin’s (1995) criteria for assessing the ‘seriousness’ of each condition are also useful for this judgement.

Currently, prenatal testing for DS is morally defensible. Future tests are likely to be diagnostic and non-invasive, and allow abortion earlier in pregnancy. These tests will likely have increased uptake rates and result in less newborns with DS. Because women do not have any obligation to support the numbers of those with DS in our community, and because abortion earlier in pregnancy is less harmful, allowing more women reasonable control over their pregnancy should be allowed, and these tests permitted.
Chapter 6  A SUMMARY: NOTES FOR POLICY

This section contains suggestions for policymakers. These suggestions are specifically aimed at testing for DS, though may prove helpful for informing testing for other congenital conditions. A separate section is devoted to current policy in New Zealand. Lastly, this chapter considers ways in which prenatal testing could be restricted, and forecasts possible harms from each approach.

6.1 Recommendations for current prenatal testing

The use of prenatal testing for DS is an acceptable expression of reproductive autonomy by pregnant women. It is acceptable, in part, because it is morally permissible to seek an abortion in the case of a DS-affected fetus. Abortion in the case of DS is at least as morally permissible as that of a fetus aborted for most ‘social’ reasons, due to the limitation of life options that DS ultimately entails. Selective abortion may be preferable for parents, either because they believe it will be more difficult for the child to live a good life, or because they want to ensure an open future for their child. These are both acceptable reasons to terminate a DS pregnancy.

That a child with DS requires more care for upbringing than a child without DS is also a valid reason for a woman to choose selective abortion. Thus, the ‘right to life’ of the fetus with DS is overruled by our regard for the mother’s reproductive autonomy. Some women may not feel this way, but will choose testing for the preparation of a child with DS. This is also a morally justifiable use of autonomy.

I have not attempted to define until what week of pregnancy abortion for DS should be permitted, but suggest that it should be the same or slightly after that for an abortion for social reasons. Prenatal testing should be offered in all weeks up to the cut-off for abortion, if this fits within the practical limits of such tests. Under a gradualist approach to fetal right to life, earlier testing is preferable to later testing as it allows earlier abortion. Prenatal testing when abortion is not an option (for example late pregnancy) is permissible in order to assist parents in their preparation for birth, but this should be weighed against the risk of spontaneous miscarriage.

It is preferable that prenatal testing is available to all pregnant women who request it, as all women are at risk of bearing a child with significant congenital disorder.
Furthermore, some women will regard testing as more desirable than others, regardless of their age. If prenatal testing is available to all women, all should be informed of its availability. Hence, the offer of testing by which one means the information that it is available – to all women is just. The fact that testing has the capacity to become ‘normalised’ should not preclude it from being offered to women. Arguably, prenatal testing is less likely to be normalised than ultrasound. This is because ultrasound has the added capacity for visualisation and direct emotional connection, which for both parents can mark a moment of ‘first contact’. Problematically, ultrasound can appear as an anticipated event to discuss with family and friends, instead of a screening procedure for congenital abnormality.

Prenatal testing for fetal conditions is fundamentally different to other aspects of prenatal care, such as blood counts and measuring fetal growth. Prenatal testing for congenital conditions is not aimed at continuing the pregnancy or ensuring the health of the fetus or mother, but serves to give information that may lead to pregnancy cessation. It is true that DS screening acts to prevent harm to both fetus and mother by avoiding unnecessary procedures, particularly on a population basis. However, it must be remembered that these are benefits which are relative to other types of test, and confer no direct medical benefit to either mother or fetus. That prenatal testing for DS differs from routine pregnancy care should be emphasized, with particular emphasis on the decision being the woman’s own, and that a range of decisions are morally acceptable. This will help limit the power of ‘routinisation’ which may bias a woman’s decision regarding testing.

Policymakers should be aware that many of the structures and processes that surround prenatal testing can be interpreted as expressing discriminatory attitudes towards those with the condition in question, and disability as a whole. However, this interpretation is not necessarily valid, and often infers negative judgements of a greater magnitude than are actually present. The interpretation that prenatal testing for DS prevents the existence of people ‘like them’ is unhelpful as it emphasises far greater similarity among people with DS than actually exists. The focus should be on human individuality and difference. Consequently, those with DS should not be labelled as a social group.
Though the ‘expressivist’ argument has some validity due to the identity-forming nature of DS, it should not preclude women’s access to the use of testing or abortion. Merely preventing testing does not mean discriminatory messages will not be expressed in other ways. Nor does the hurt conferred by such messages outweigh the ethical benefits conferred by an increase in a woman’s reproductive autonomy. Nevertheless, the expressivist argument is helpful in that it forces us to reflect on how the offer of prenatal testing is presented to women, and our treatment of those with disability in our society in general. We can reduce the hurt felt by those opposed to prenatal testing by ensuring that the needs of people with disabilities are met, and that are not subject to stigma. Suggested avenues for action include laws that outlaw discrimination against the disabled and encourage increased contact, knowledge and exposure of people with disabilities’ lives. Some steps such as the United Nations Convention for the Rights of Persons with Disabilities have been taken, and should be applauded.

With these safeguards in place, prenatal testing for DS is a morally defensible procedure that focuses on facilitating individual choice. It is therefore significantly different to the eugenic practices of the early 20th century (see Afterword), which neglected the value of individual autonomy in deference to the ideals of the state. Uncompromised choice enables the individual to decide whether a child with DS is compatible with a woman’s idea of ‘the good life’. This choice should be made by the woman in question, and this is what currently occurs.

It is reasonable that public money is allocated to fund DS screening and diagnosis. Though these tests are different from other antenatal tests, they provide information that is significant for a large number of women. An ideal screening/testing framework allows the woman to give fully informed consent. A screening programme which actively aims to reduce the number of infants with DS is unlikely to facilitate free choice; as pressuring women to abort will help it achieve its aim. Therefore reducing the numbers of infants with DS should not be an aim of a screening programme, although it may be a consequence.

It should be noted that weighing the cost of a screening programme against the savings conferred by the prevention of DS births tells us nothing about whether screening or diagnosis for DS is justified. However, it may inform us whether a testing programme is *economically viable*. Economic considerations inform nearly all policy
decisions, and while healthcare operates under limited resources evaluating the overall cost of a screening programme will be continue to be necessary.

All disorders, be they congenital or acquired, have a financial cost attributed to them, and DS is no exception. Assessing the cost of a life with DS with its ongoing healthcare and support requirements is no different to assessing the cost of cancer treatment, or injury prevention. This cannot be used to label people with DS as a ‘burden on society’ any more than people with any other condition. Indeed, many other disorders are far more costly to treat. The ways in which those with DS and other conditions contribute to the lives of their friends, families and communities are unquantifiable. Human beings and the relationships they sustain cannot be considered burdens in the way that cost-benefit analysis predicts. If we accept this, then we can hold two mutually compatible views; that those with DS require additional expenditure for their needs to be met, and that they are not burdens on society.

If a screening programme results in less neonates with DS then some money will be ‘saved’ in terms of healthcare and social welfare. But maximizing these ‘savings’ in a screening programme is untenable as it would require some form of coercion, and this would make the process morally unjustified. However, projecting the ‘savings’ from the success of a voluntary, ethically sound screening programme is defensible, if this is a by-product of facilitating parental choice. In short, appreciating that some economic gains will occur as a consequence of an ethically sound screening programme is acceptable policymaking, so long as optimizing these ‘savings’ does not become the aim.

That widespread prenatal testing may result in a reduction in the number of people with DS does not outweigh the moral gains conferred by allowing all women access to tests. Whether a loss in numbers of those with disability results in a loss of support is uncertain, and it is more likely that more resources will be available to those in need. It should be noted that there is no need to retain the exact distribution of any particular trait in society, and it is unlikely that DS will vanish. As I have written previously (Cole & Jones, 2013), a reduction in the number of individuals with DS does not mean that we will value them any less than in the past. Indeed, reduced numbers of people with DS could also arise from the use of a ‘cure’. But in this case, treatment would not be withheld. An analogous argument can be made to support prenatal testing.
The most pressing area of policy in need of change is the information on DS which is provided to pregnant women. This information has for too long been excessively medically oriented, and sometimes even inaccurate. Women are not making a medical decision but a holistic one, and although future health status is a relevant consideration it is by no means the only one. Thus, women need to be informed about people with DS’s lives, their interaction with family and friends, and what it is like to raise a child with DS. Further research is needed to find the answers to these questions. Some information is now available (see Chapters 1.4 and 1.5), though past studies are limited by low response rates. Ideally, the main messages from these studies should be distributed to all those faced with the choice of DS screening. Informed consent is paramount in beginning of life decisions as it is in end of life decisions, because the possible outcomes are so dramatic.

Another important area to be addressed is the education of medical professionals. Though the view that the medical profession is dismissive and uncaring towards those with disability is likely a gross exaggeration, it is probable that some negative attitudes remain. This is unsurprising, as these are prevalent among the general public to an extent; but this does not justify them. More efforts should be made in medical curricula to include the stories and experiences of those with disabilities, and not least, the experiences of those with DS. Particular emphasis should be placed on reiterating that it is the social stigma and lack of support which contribute most to the negative experience of living with disability.

6.2 Recommendations for NIPD and PGD

With the present in mind, policymakers should pay regard to what lies over the horizon. Tests such as non-invasive prenatal diagnosis (NIPD) fit entirely within the existing ethical paradigm; that is, they do not raise significantly dissimilar moral questions to the current tests in use. However, NIPD makes addressing some issues more pressing. Informed consent could be difficult to obtain, with women ignoring the potential implications of NIPD and regarding it as ‘just a blood test’ (de Jong et al., 2009). Women may feel less justified in refusing diagnosis, due to the absence of spontaneous miscarriage risk (Newson, 2008). Because the test carries little or no risk of physical complications, routinisation and even social pressure to have the test could be an issue.
(Chitty et al., 2012; Kelly & Farrimond, 2012). This would be more concerning than the routinisation of screening, due to these tests’ rapid diagnostic power.

However, policymakers should be aware that the potential benefits of NIPD outweigh the risks (Cole & Jones, 2013). The most important advantage is the removal of the risk of fetal loss associated with invasive techniques, a consideration which is often the most important factor for women considering a prenatal test (Chitty et al., 2012). That test results could be available earlier in pregnancy is preferable under the gradualist approach to fetal right to life. That a blood test has such diagnostic power is acceptable if women are adequately informed and reflect on this prior to deciding whether or not to be tested. Thus, the most important challenge of NIPD is to strengthen existing avenues for counselling, and to enable informed decision making. Continuing to offer screening before NIPD may have several benefits, including ensuring informed consent and minimizing social pressure to take the test. It is of significance that NIPD is currently not diagnostic, and at the moment should only be considered as a very accurate screening test. However, this is likely to change. Policymakers should continue to monitor the development of NIPD, as this is a constantly developing field.

Preimplantation genetic diagnosis (PGD) for DS has been permitted for a number of years in New Zealand (National Ethics Committee on Assisted Human Reproduction, 2005) and worldwide. Like prenatal testing, PGD is another form of genetic selection, and thus similar objections apply (such as devaluing the lives of those with disabilities, and routinisation). But with PGD these objections carry less weight, as choosing a specific embryo to avoid a child with DS is a much lesser action (and constitutes less harm) than the rejection of a wanted pregnancy. If we hold that prenatal screening and diagnosis for DS is justifiable, we should do the same for PGD.

6.3 Recommendations for New Zealand

The National Screening Unit (NSU) is a section of the Ministry of Health which monitors and is responsible for all screening programmes in New Zealand. It recommends that “all pregnant women are offered information about screening for Down syndrome and other conditions, preferably in the first trimester of pregnancy” (National Screening Unit, 2011). Instead of a direct offer this is now presented as ‘advice of its availability’ due to work by Saving Downs and other groups. This allows some
practitioners to “exercise their right of conscientious objection” and not provide the service themselves (Saving Downs, 2012a).

I have argued that all women should be advised of the availability of prenatal screening, and I have previously commented on the dangers of the routinisation. If changing the ‘offer’ to ‘advice of availability’ helps to mitigate this risk, then this is a step in the right direction. However, I would be concerned if a practitioner objected to providing the service of prenatal screening. Such a practitioner is not providing a service of relevance to a significant number of women. Though prenatal testing is ethically problematic, it is morally justified on the whole. I would suggest that this practitioner would not be providing the highest standard of care to his/her patients.

With that in mind, I would not go so far as to withdraw that practitioner’s ‘right to conscientious objection’ unless it was precluding women access to prenatal testing. Such a scenario could be foreseen if he/she was the only maternity carer in an isolated rural town. In these cases, such a ‘right’ should be overruled, as it harms some women who may benefit from testing. My argument is not unlike that which opposes the ‘right’ of pharmacists who refuse to supply emergency contraception (Cantor & Baum, 2004).

Further action by Saving Downs has resulted in a change in the pamphlets given to patients who are offered screening (Saving Downs, 2012a). These steps are to be supported, if these resources realistically describe the value of living and raising a child with DS, instead of simply concentrating on associated medical conditions and impairment. The abortion of a fetus with DS is a holistic decision, not only a medical one; information given to those faced with testing or abortion should be balanced, to reflect this.

There have been some proposals that all those faced with receiving a positive result from testing should be referred to a family member of a child with DS (Saving Downs, 2011b). Such a policy may not be logistically feasible as there are many positive diagnoses of DS every year. Even if viable, this process could still remain problematic. The family member may well have an experience of living with or raising a child with DS which is non-representative of that of most families. Indeed, such is the variability of DS that the child they have raised could have very different limitations to that carried by the pregnant woman. The opinion expressed by the family member is likely to be subjective, and any counselling or advice is unlikely to be non-directive. With these
factors in mind, I suggest that a trained practitioner is most likely to be able to give objective information most in line with what will be helpful for women, and would be a more suitable referral. That said, women should continue to be provided with information about organisations such as the New Zealand Down Syndrome Association which may offer these services.

A change desired by Saving Downs (2012c) is to suggest that DS does not classify as a serious fetal physical or mental handicap [sic] as required by New Zealand law (Crimes Act 1961). Though DS does exhibit variability in the limitations it confers, all with DS exhibit some degree of significant cognitive impairment. Those with DS are also at vastly increased risk of multiple physical malformations, though these cannot be picked up by testing alone. These features are medically serious, and significantly influence the life prospects of the individual with DS (and, for some women, will be considered ‘holistically serious’). Because of this, DS should continue to be regarded as a serious congenital condition and should remain grounds for abortion under New Zealand law. It should be noted that even were Saving Downs’ proposed change to be accepted, DS would probably still be grounds for abortion in New Zealand through the pathway currently pursued for ‘social’ reasons (via women asserting that psychological harm would come to them through the bearing of the child).

I have one recommendation for the regulation of PGD. In New Zealand, PGD for DS is permitted when “the woman is of an advanced reproductive age” or when “the woman has had recurrent implantation failure or recurrent miscarriage” (National Ethics Committee on Assisted Human Reproduction, 2005, p. 5). I suggest that these guidelines should be removed, and PGD for DS should be permitted for all women undergoing in-vitro fertilisation (IVF). This is in line with current DS screening practice which allows all women access to DS screening, and by proxy all women access to diagnosis and termination. This does not mean that all women undergoing IVF should routinely be offered PGD for DS, merely that women should not be legally barred from doing so. A similar argument would apply to other conditions tested for with DS screening and diagnosis in New Zealand, such as trisomy 13 and 18, and other metabolic disorders.
6.4 Recommendations for the prenatal testing of other congenital conditions

I am concerned that policy decisions regarding the testing of other congenital conditions may be solely influenced by medical severity. Though this consideration is important, it should not be the only one. I urge policymakers to take into account how the condition affects people’s lives; both the lives of the parents, and that of the resulting child. By assessing the whether caring for a child with a condition significantly disrupts parent’s lives, and whether the condition significantly limits a child’s open future, we may be better informed of which conditions should and should not be able to be tested.

6.5 Restricting prenatal testing: Consequences and harm

It is of note that there are many movements which aim to prohibit the wide use of testing for DS (Saving Downs, 2011a), not least the anti-abortion lobby in the US (King, 2012). Though I believe a woman’s right to choose whether to bear a child is undeniable up until around 20-24 weeks of pregnancy, as explained in Chapter 3 this right is stronger when the fetus has DS. If some jurisdictions choose to restrict abortion, it should be noted that selective abortions for DS are frequently more permissible than social abortions. The same may be true for other congenital disorders which result in considerable impairment.

There are several ways that prenatal testing for DS could be limited. Public subsidies could be removed from tests; this would result in those with less income being unable to access testing. Testing could be restricted to only those over a certain age; but women of all ages are at risk of a DS pregnancy. Though younger women have reduced risk, some may regard any risk as of great significance. Women of ages either side of this arbitrary cut-off would be almost identical in risk, but vastly different in the options available to them. An alternative is that testing could be completely prohibited; to do so would be to deprive all women of an important and justifiable test, not to mention older pregnant women who are at high risk of aneuploidy possibly avoiding conception altogether. All of these outcomes are undesirable.

DS can still be detected via ultrasound, a procedure which many women routinely receive several times as part of routine pregnancy care. If ultrasound was still permitted but DS testing prohibited, DS would still be detected in many pregnancies.
Ultrasound is less specific than screening or invasive diagnosis, and if abortion was to be chosen there would be a significant risk that the fetus did not have DS. In short, many pregnancies would be terminated unnecessarily.

All of the current prenatal tests for DS (ultrasound, DS screening, invasive diagnosis) also detect other congenital conditions, such as trisomy 13 and 18, and various metabolic disorders. It is unlikely that testing for these disorders would be banned, as they have a very poor quality of life and a life expectancy of no more than a few months. Indeed, it seems inappropriately harmful to outlaw tests for conditions where the child has a painfully short life, which can cause much distress and anguish for parents. Therefore, tests with the ability to detect DS will continue to be used. But if these tests are still permitted and DS is detected in the fetus how then would policy proceed?

One position to adopt is similar to the New Zealand Down Syndrome Association position, which “does not consider Down syndrome is in itself a reason for abortion” (New Zealand Down Syndrome Association, 2012, p. 6). Whether this position has any normative force is doubtful; as stated in Chapter 3.2.1 and 4.1.1, women do not abort just because of DS, but reject what parenthood of a child with DS entails. This is a pluralistic, holistic decision, based on many circumstances in the woman’s life and complex perspectives on the nature of DS (Bryant et al., 2006; Garcia et al., 2009; Raz, 2004). Thus the obvious riposte is that already, no abortions take place purely because of DS; abortions occur because of how DS fits in with the woman’s projected life plan, encompassing many different values.

A clearer perspective is held by anti-testing lobby group Saving Downs, which advocates a “repeal of ‘foetal abnormality’ as a grounds [sic] for abortion” (Saving Downs, 2012c). This position would entail a significant restriction of a woman’s rightful expression of reproductive autonomy, as abortion in the case of DS is justified. Indeed, the Saving Downs position against any fetal abnormality is even harder to defend; for disorders such as anencephaly (where the newborn will die within two weeks) would not be sufficient reason for abortion. Not to allow such an option seems unjustifiably harmful to the parents of such a short life, and, arguably, the child itself.
For these reasons it is undesirable to restrict prenatal testing for DS, and untenable to prohibit it. While some change to policy is needed, the current process of testing for DS is morally defensible.
Concluding thoughts

Continually, the role of the doctor seems to change. Though at one time solely focused on treating to cure, members of the medical profession are increasingly being asked to provide services where there are no correct or obvious answers.

Reproductive technologies are driving this change. The urge to have a child is sometimes described as the strongest urge that a person can have. Because this urge is so strong, and because the child is theirs, some parents mistake this urge as a requirement that the resulting child must be just like them. Parenthood is not about ‘raising another one of you’. It is about giving another the chance to grow, with care and direction, to find him or herself.

This is the context within which prenatal testing should be approached. Prenatal testing for DS is not a test which allows children to be identical to their parents. Rather, it can be used by parents to allow open futures for any resulting children. This does not necessarily advocate for genetic enhancement; action in this way is limited to other considerations, such as harm and equality. In any case, I suggest that parents using prenatal testing to ensure open futures are not undermining the tenets of parenthood; if anything, they are upholding them.

We live in an age of medicalisation. Health-related information which was once confined to dusty medical shelves is now available through the click of a button. ‘Home diagnosis’ using the internet is increasingly problematic, while television shows set in hospitals become increasingly popular. Anything which could be a symptom must have a cause. Natural processes, such as pregnancy and ageing, are seen as inherently risky, requiring the utmost in supervision. Natural emotions, such as ‘sadness’, are quickly classed as depression.

The doctor must be aware of these processes. A doctor’s role is often to treat, but always to care. A pregnant woman faced with a positive diagnosis of DS cannot be treated in a conventional sense, but aiding her to reach an informed decision is caring for her in ways in which many others cannot. Stopping her from over-medicalising her pregnancy is part of this duty. Helping her recognize that choosing whether or not to raise a child with DS is not solely a medical decision but a holistic one is of the utmost importance. Yet it is easy to forget, when discussion takes place in a workplace
surrounded by appointments, procedures and medical equipment. And it is easy to ignore, when the limitations of a medical approach are so often overlooked.

New tests will be developed, and new challenges will abound. Conditions thought forever undetectable may be in mainstream use in 10 or 20 years. Doctors should not only care for us, but we should care for them. By making sure that the tests available are morally justified, doctors and other maternity carers can be secure in the testing that they offer. Though the case of DS is particularly divisive, other disorders may be more so. Forecasting is helpful, as tests can move from laboratory to clinic with incredible rapidity. Above all, ongoing reflection is needed; to ensure that our treatment of pregnancy follows a path that will enhance, rather than detract, from our humanity.
Afterword: On the subject of eugenics

This thesis has not addressed the subject of ‘eugenics’; namely, whether testing for DS is or is not eugenic. Though this question is relevant to the question of whether or not testing is justified, I did not feel that its placement was necessary in the main body of the text. What follows is a brief overview of eugenics, and a justification for its omission.

That a procedure is ‘eugenic’ is a common objection to any process which interferes with natural reproduction. The word ‘eugenic’ itself means “pertaining or adapted to the production of fine offspring” (Oxford English Dictionary); in simpler terms, “well born” (Mahowald, 2002, p. 223). It was coined by biostatistician and human geneticist Francis Galton (1883) in his widely influential text “Inquiries into human faculty and its development”. Galton and other eugenicists built on the work of statisticians who used statistics to describe and categorise bodily phenomena, placing them along a strata of distribution to exhibit ‘standard’ and ‘non-standard’ populations (Davis, 1995). Eugenicists were further influenced by Charles Darwin’s theories of evolution and natural selection.

Galton and other eugenicists felt that selective breeding for the human race had the potential to produce a wealth of benefits; at the least, “a galaxy of genius” (Galton, 1865, pp. 156-157). Some authors have distinguished between ‘positive’ and ‘negative’ eugenics. Positive eugenics encourage reproduction between those with ‘desirable’ characteristics, whilst negative eugenics discourages those with ‘undesirable’ characteristics from reproducing (Kevles, 1985). Most of Galton’s suggested eugenic policies were variations of positive eugenics, such as tax schemes which encouraged intelligent people to marry each other and have large families (Cowan, 2004). However, most people today regard the negative eugenic practices of the 20th century as the prime example of eugenics.

Eugenics as a political movement gained ground throughout the early 20th century, and by the 1920s eugenic movements existed all around the world (Kevles, 1999). This movement translated into policy, with 24 American states together with Canada and Sweden possessing sterilization laws by the late 1920s (Kerruish, 2000). By the mid-1930s in the US more than 20,000 sterilizations had been legally performed. The most severe use of negative eugenics occurred in Nazi Germany, where 225,000
people labelled as ‘feebleminded’ were sterilized (Kerruish, 2000) and thousands of ‘defective types’ - the intellectually and physically impaired - put to death. By the end of the regime, 5.6 million Jews and 250,000 gypsies had been killed in the pursuit of ‘racial hygiene’ (Kevles, 1985).

When this was discovered to be the stark extreme of eugenic thought, the practice of eugenics rapidly lost popularity (Buchanan et al., 2000). With better understanding of the relationships between genetics and the role of the environment in shaping one’s phenotype, much of the eugenicist philosophy has since fallen out of scientific, political, and philosophical favour.

**Is testing for Down syndrome eugenic, and does it matter?**

Some, such as Rita Joseph (2012) and the anti-testing group Saving Downs (Saving Downs, 2012b) assert that testing for DS is a eugenic practice. Their main justification for this is their objection to selective abortion (Joseph, 2012). They contend that because a process is eugenic, it *must* be opposed. This approach aims to link all eugenic processes with the worst excesses of the Nazi Regime. For these objectors, prenatal testing is a ‘slippery slope’, which if we continue to follow will result in a Third-Reich style dystopia.

This approach is inaccurate, reactionary and sensationalist, frequently catching the attention of the media (Ashby-Coventry, 2012). One of its most telling flaws is its narrow conception of what eugenics really is. If eugenics means the forced euthanasia of thousands of the intellectually and physically impaired, then eugenics is well and truly dead. More accurately, if eugenics means to try to produce ‘fine’ offspring, then it is surely alive; for we are all eugenicists, as we choose with whom to mate and reproduce. Any form of prenatal care is eugenic, as it aims to protect the health of the fetus, thus ensuring a healthy newborn. Pregnant women who avoid certain foods, alcohol or reduced alcohol intake can all be classed as eugenicists. But these actions are usually seen as responsible, and morally praiseworthy. Eugenic actions can take many forms (Paul, 1994); these are not necessarily good or bad. Because of this, merely labelling a practice ‘eugenic’ does not tell us if it is justified (Mahowald, 2002).

Mary Mahowald (2007) has tried to distinguish between ‘good’ and ‘bad’ eugenics. Using her methodology, ‘good’ eugenics facilitates autonomous decisions by
potential parents; whereas ‘bad’ eugenics is state enforced, leaving no room for individual choice, and directed at the avoidance and reduction of specific traits. For the charge of ‘bad eugenics’ to hold, DS testing would have to meet some or all of these criteria.

It is no accident that these questions have all been addressed in the preceding chapters. Currently, women are given a choice whether or not to test for DS; it is not required by any state. Some have suggested the presence of ‘institutionalised eugenics’ via coercion by the health workforce (Dixon, 2008), reducing rather than increasing women’s autonomy (Klein, 2011). As examined in Chapter 5.1 and 5.2, evidence suggests this happens only in a few cases, and disregards the options that testing provides for many pregnant women. Some express dissatisfaction at the prenatal testing of specific traits (Asch, 2000). As examined in Chapters 3.2 and 4.1, the decision to have a selective abortion for a specific trait encompasses many variables, and need not (and often does not) necessarily reflect discriminatory attitudes towards people with disabilities.

Some worry that though a woman’s autonomy is preserved via the current process, collective individual choices will still bring population change; in other words, a ‘eugenics through the back door’ (Duster, 2003). This issue has been discussed in Chapter 5.3, with the conclusion that though population change may occur from testing, this does not outweigh the moral gains for a woman to access prenatal testing.

Mahowald’s (2007) approach is helpful in that it alerts us to features of reproductive technologies which could be morally problematic. But the categorization of ‘eugenics’ in this way tells us nothing especially new, as ‘eugenics’ is not inherently bad. Why have ‘bad eugenics’, and not ‘bad reproductive procedures’; or just ‘bad’ on its own? The label of eugenics is redundant in informing our viewpoint on a procedure, as it tells us nothing new except that it is involved in some aspect of reproduction.

It is much better to assess the issues at play in the use of any reproductive technology. For prenatal testing for DS, this has been among others, the moral permissibility of abortion, the argument from expressivism, and the problems associated with offering prenatal testing. Through this approach, the question of whether testing for DS constitutes ‘bad eugenics’ has already been answered. The answer, at least in the present context, is a resounding ‘no’.
That we are aware of the eugenic excesses of the past helps us prevent them from occurring again. Movements which require the sole precedence of one ethic (in this case, the improvement of the human gene pool) over all others (such as respect, prevention of harm, or individual autonomy) will often result in misery and injustice. This thesis asserts that DS testing is eugenic, in the same way that avoiding alcohol in pregnancy is eugenic. However, labelling something as ‘eugenic’, in effect, tells us nothing new.
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Appendix

Testing times: Do new prenatal tests signal the end of Down syndrome?

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Abstract

Since 2010, prenatal screening for Down syndrome (DS) has been offered to all pregnant women in New Zealand. The programme has been criticized by several groups, on claims that screening is eugenic and discriminatory towards those with DS. Recently, tests have been developed that may one day prove more efficient than current screening methods. They are an example of ‘Non-Invasive Prenatal Diagnosis’ (NIPD), which enables diagnosis earlier in pregnancy with less risk of complications. If the current programme raises objections, what threats does this new and seemingly more attractive technology pose to the DS community?

We argue that NIPD is simply an extension of current screening methods, raising similar ethical concerns. Presently, the programme shows little evidence of ‘eugenics’, demonstrated by moderate uptake rates and varying attitudes towards disability. We do not regard the offer of screening to be threatening, as women choose whether or not to be screened depending on their own personal circumstances. One day, prenatal testing may result in fewer people with DS; but past and present trends indicate these individuals will continue to be supported, irrespective of ‘group size’. Care and respect for the disabled will remain essential, regardless of a woman's decision over her pregnancy.
Testing for Down syndrome

In October 2011 a new prenatal test for Down syndrome (DS) was unveiled in the United States. The test analyses proportions of fetal DNA in maternal plasma to give a risk estimate of DS in the fetus. This is an example of ‘Non-Invasive Prenatal Diagnosis’ (NIPD), which can also detect Trisomies 13 and 18. Though not specific enough at present, with improvement NIPD could make current prenatal screening and diagnostic methods for DS unnecessary. However, groups such as ‘savingdowns.com’ believe that New Zealand’s current screening process is ‘eugenic’, discriminating against those with DS by ‘the prevention of their births’. Further concerns are raised over these new tests, since NIPD has the potential to be used in more pregnancies than with existing screening. Therefore it is timely to ask: what would NIPD mean for the DS community? Furthermore, what does testing, both now and in the future, say about our attitudes towards DS, and disability as a whole?

Since 1968 women have been selectively offered prenatal diagnosis for DS on the basis of advanced maternal age. This ‘opportunistic’ method of screening was declared ‘unsafe and should not continue’, as unnecessary numbers of invasive diagnostic procedures were harming some pregnancies. In 2010 ‘quality improvements’ were introduced which resulted in New Zealand’s current screening programme, which tests for DS and other congenital conditions. The method of screening differs, depending on gestational age. The majority of screening tests are taken before 14 weeks of pregnancy, via the ‘First Trimester Combined Screening’ pathway. Here, information on the mother (such as maternal age and smoking status) is combined with a maternal blood test of two serum markers and the results of a Nuchal Translucency (NT) test on ultrasound. After 14 weeks’ gestation ‘Second Trimester Maternal Serum Screening’ is offered, which utilises information on the mother and the results of four serum markers, without a NT scan. Both of these screening pathways give a numerical risk estimate of congenital malformation in the fetus, which is then conveyed to the patient as either ‘low’ or ‘increased’ risk. Throughout the process women are to be reminded that the screening is voluntary, that they can opt out at any time, and that partners and family can be involved in decision-making.

Currently, just over half of all pregnancies receive DS screening, though it is offered to all pregnant women. Thus there is considerable room for uptake to increase,
which may well result from the use of a quick, safe test such as NIPD. The ‘quality improvements’ of 2010 were developed to provide equality of access and safety for mother and fetus, and NIPD could prove to further fulfil these aims.

**Non-Invasive Prenatal Diagnosis**

In the case of an ‘increased risk’ result, two diagnostic techniques can be used to confirm DS (and other genetic abnormalities) in the developing fetus. Chorionic villus sampling (CVS) is used earlier than 14 weeks of gestation, and amniocentesis is used after this time. Both procedures carry with them a spontaneous abortion risk of around 1%. With current screening most pregnancies subjected to CVS or amniocentesis do not actually have a DS fetus, and as a result, fetuses are lost as a consequence of these diagnostic procedures. The primary advantage of NIPD is that there is no risk of spontaneous abortion, because diagnosis is based on only a blood sample.

Currently, NIPD has the potential to be used from 10 weeks of pregnancy, similar to CVS and earlier than amniocentesis. In the future, NIPD could be used from seven to nine weeks, as fetal DNA is found in maternal blood at a very early gestational age. Results could be obtained more quickly, as current invasive procedures have turnaround times of one to three weeks. At present diagnostic results are rarely received before 12 weeks, and often after 17 weeks of gestation, leading to late terminations which can be traumatic and (at times) dangerous. An earlier diagnosis would allow women more time for decision-making, and the option of an earlier, safer termination with less emotional and mental repercussions.

It has been reported that women express interest in NIPD, primarily due to the absence of risk of spontaneous abortion. Some women find diagnosis helpful to prepare for the birth of a child with DS, and NIPD would be preferable to amniocentesis/CVS as there are no major complications. NIPD may prove more attractive for district health boards, by reducing the number of costly invasive procedures at specialist care centres. For these reasons, NIPD is likely to be used in more pregnancies than current diagnostic procedures at some stage in the future. Should costs drop and clinical efficacy be proven, NIPD could eventually make current screening methods redundant as well.
But what would NIPD mean for the DS community? Increased uptake of tests will result in increased detection of DS, and probably more terminations. The number of DS births may, as a result, drop. However, it is unlikely that DS will disappear. Abnormalities escape detection using even the most rigorous diagnostic techniques, and there will always be women who do not wish to undergo testing.

But as more pregnancies are tested, will DS become a 'rare' disorder? In time, perhaps. However, the life expectancy of those with DS is increasing, and is likely to soon approximate that of the non-DS population. This will mask, at least temporarily, any effect of NIPD on the prevalence of DS. Hence, even with a rapid increase in the uptake of NIPD, it is unlikely that the numbers of those with DS will change markedly in the near future.

NIPD and current DS screening tests both provide comparable information, and enable similar choices for pregnant women. Because of this, the ethical issues likely to be raised by NIPD will be analogous to those associated with current screening. 'Savingdowns.com', an anti-screening group, argues that a nationwide DS screening programme is simply a money saving exercise, initiated by a government which views individuals with DS as nothing more than a drain on society. 'Savingdowns.com' claims that the current screening costs '$75,000 per [terminated] child with Down syndrome'. However, such wording misrepresents what screening provides to the majority of women; namely, reassurance in the case of a 'low risk' result. To evaluate DS screening on the basis of cost-effectiveness is to compare DS to other screened, treatable diseases, such as breast and cervical cancer. In these cases, a 'cure' is the overall aim. There is no 'cure' for a DS pregnancy, indeed this is not the aim of the test. The test's purpose is to give women information on the pregnancy, not prevent a DS birth. The value of this knowledge to women is impossible to quantify, because the choices it makes possible would otherwise be unavailable. For these reasons cost-effectiveness should not be used to assess DS screening, despite claims that it is the overall aim.

Is screening for Down syndrome 'eugenic'?

Anti-screening groups frequently label the current screening 'eugenic' in nature, making distinct comparisons to the killing of the disabled in Nazi Germany in the 1930s-40s. This clouds the debate around the ethics involved, since current screening is voluntary, not state-enforced. Patients are given a choice whether or not to be screened,
and how to respond to the resulting information. DS screening does not serve to systematically erase the congenitally disabled from the population; it provides information for patients about their pregnancy. This is not the start of a ‘slippery slope’ to Third Reich genocidal acts; as shown below, support and advocacy for the disabled has never been greater. Nevertheless, it is important to ensure that women are never coerced into accepting screening or subsequent termination, and a decision is made which is appropriate for them.

If all pregnant women were persuaded to be screened and unable to make voluntary decisions then eugenic overtones would indeed be present. Such persuasion does not need to be administered by the state; if severe pressure was exerted by health practitioners a form of ‘institutionalised eugenics’ could still eventuate. Such a practice would require all women, or a large proportion of them, to be coerced in the same way. However, with only 55% of pregnancies utilising DS screening,7 this does not appear to be the case. Thus to argue that the screening programme is ‘eugenic’ seems inaccurate, as nearly half of all pregnant women are declining the offer of screening. This points to the success of fair, supported choices free from state or medical coercion.

Accusations of ‘institutionalised eugenics’ are better directed at termination of pregnancy, as 90% of women who receive a positive result from CVS/amniocentesis proceed with termination.17 This number seems high until placed in context: those unlikely to consider termination on the grounds of DS often decline diagnostic testing. If there remains a suspicion of ‘institutionalised eugenics’, it is unlikely that any health workforce could unilaterally enforce such a decision on an entire population, for several reasons. Those in the health workforce are far from homogeneous, and have varying perceptions of disability. No longer do patients leave their choices purely in the hands of doctors, while a patient’s right to refuse medical treatment is well-recognised and enshrined in law. Lastly, non-directive counselling has been shown to be beneficial in allowing women to make fair, independent decisions for screening decisions,18,19 and is offered to women both before and after diagnostic testing.6

There is evidence that a minority of practitioners may attempt to emphasize negative aspects of DS so parents will favour termination of pregnancy; however, a similar number emphasizes positive aspects to encourage pregnancy continuation.20 Hence, to argue that the current programme is eugenic is an over-generalization, even
though some health professionals may be unduly persuasive in offering termination. This fault is not implicit within the screening process per se; it points to a flaw in the education of health professionals, where a proportion are inadequately informed about the quality of life of those with DS. While DS results in varying levels of intellectual disability, those affected report a consistently high satisfaction with their lives. The vast majority of persons with DS feel that they are capable and have self-worth, and love their families and friends. Children with DS are frequently described as being more content, caring and loving than non-DS children. It is, therefore, incumbent upon obstetricians, GPs and midwives to ensure that information such as this is conveyed to women involved in the screening pathway. With tests such as NIPD likely to be used earlier and more frequently in pregnancy, this becomes increasingly important.

**Supporting individuals with Down syndrome**

But does the mere offering of a test not subtly imply that DS is undesirable, a ‘disease’ best avoided? We argue that this is not necessarily the case, although we recognise that children with DS require more care and support than other children. This support is often required throughout life as in most cases a person with DS cannot live fully independently. For some families, raising a child with DS will be immensely difficult, and so by offering termination we concede that raising a DS child will be seen as being too great a burden for some. This is similar to the offer of termination for other serious congenital disabilities, a situation that has prevailed since the 1978 amendment of the Crimes Act in New Zealand. Society offers a choice, not a routine procedure; it is the woman, not the state, who makes this judgement. The assumption is that they make this choice in regard to their own life circumstances, and not merely because a screening process is offered within the first 20 weeks of pregnancy.

As a consequence of the availability of NIPD and any further tests that may be developed in the future the numbers of those with DS may fall. However, there is no indication that society will cease to value these groups, even though they number less than in the past. Disabled persons gained recognition and respect throughout the 20th century, regardless of their group size. Awareness and services for the disabled have grown dramatically and funding is set to increase for the near future. Some argue that a reduction in number of those with a disability like DS will reduce the standing, recognition and support of such individuals in society. This is unsubstantiated, since
there is no evidence that society neglects to treat rare disorders because there are few with the condition. For instance, we do not value and support those with DS more than those with Fragile X syndrome, on the grounds that DS is more prevalent. There is no evidence that our care for those with spina bifida is inferior to that of 10 years ago because the incidence of spina bifida has decreased.\textsuperscript{27,28} We value and treat individuals as persons, supporting them in regards to their needs, not the number who shares their disorder. Along similar lines we will continue to support those with DS, and this is in no way jeopardised by women’s decisions for their own pregnancy.

We concurrently offer prenatal screening and value the disabled by upholding several values in society. First, we value an ethic that stresses the importance of ‘doing the most good’. On these grounds we accept that in some cases, the perceived disadvantages resulting from a DS pregnancy (to child and family) may outweigh the perceived good from the child’s life. Second, we value reproductive liberty, the ability to make individual decisions over one’s pregnancy. Others, such as the state, are limited in their control of this right. Alongside these we uphold dignity, respect and justice, realising that those who are disabled demand equal respect as citizens, thereby deserving support from society.\textsuperscript{29} Inevitably, these values must be held in some tension; but as long as they are recognised as important, we will make sure one (e.g. reproductive liberty) never fully undermines another (e.g. respect for the disabled). From this, we can argue for two compatible viewpoints - that screening is justified, and that the disabled will continue to receive support and respect from society.

The advent of less invasive tests such as NIPD places increasing demands upon our ethical awareness. While NIPD does not automatically lessen the value society places on disabilities such as DS, technological efficiency must never be our sole consideration in the use of such tests. It must be balanced by serious regard for continued, and, if necessary, increasing support for children and adults with these conditions. It should be noted that NIPD is still in its infancy, with technological advances permitting detection of other conditions, reducing cost, and improving specificity all required before NIPD is likely to be offered as standard care.\textsuperscript{30} Regardless, should NIPD or tests like it one day replace current screening methods, unwavering advocacy for those with disability will remain of paramount importance.
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