# CHAPTER FIVE

## Serious disease as kinds of living

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In December 2003, a Church of England curate from Chester in North West England challenged West Mercia police's failure to investigate the termination of a pregnancy in 2001 involving a 28-week old foetus which had been diagnosed with a cleft lip and palate using ultrasound technology. In England, medical termination after 24 weeks is only allowed to save the pregnant woman's life, to prevent grave permanent injury to the health of the pregnant woman or if it is judged that the future child would be 'seriously handicapped' (United Kingdom 1991).[1] The curate's charge was that this termination had in fact been an unlawful killing because there was no risk to the life or health of the woman nor was there any risk that the child would be born seriously handicapped.

Just over a year later, in March 2005, the Crown Prosecution Service informed the curate that she had lost her challenge. The issue to be determined, according to the Crown prosecutor, had been 'whether the two doctors who had authorised the termination were of the opinion, formed in good faith, that there was a substantial risk that if the child were born it would suffer from such physical and mental abnormalities as to be seriously handicapped' (Crown Prosecution Service 2005). Following a review of the patient's medical records, guidance from the Royal College of Obstetricians and

[1] Up to 24 weeks, medical termination in the UK may take place if two doctors agree that continued pregnancy would involve "risk of injury to the physical or mental health of the pregnant woman or any existing children of her family… greater than if the pregnancy were terminated" (United Kingdom 1991).

Gynaecologists (RCOG) on medical termination of pregnancy as well as evidence from a number of professionals involved in the patient's counselling and treatment and other medical experts, the Crown prosecutor concluded that 'the abortion was due to a bi-lateral cleft palate and was legally justified and procedurally correctly carried out' (EWHC 2003). He would later add, 'I consider that both doctors concluded that there was a substantial risk of abnormalities that would amount to the child being seriously handicapped' (cited in Gledhill 2005). The case, which has been debated widely in the UK (see Scott 2005a), is a helpful starting point for a chapter aiming to explore the terms and conditions that allow for normative estimations of 'good life' in reproductive medicine today.

Studies of the social, legal and ethical implications of selection practices in reproductive medicine today have in large part focused on four key areas: the problem of (non-)directive counselling (Pilnick 2002, Rapp 1988, Williams, et al. 2002b); debates about how to (and who should) define what a 'life worth living' and a 'serious disease' are and where to 'draw the line' when it comes to selective reproductive practices (Scott 2005b, Williams, et al. 2002a, Williams, et al. 2007); whether or not new reproductive technologies are a form of 'backdoor eugenics' and/or increasingly used to produce 'designer babies' (Duster 2003, Gosden 1999, Shakespeare 1998); and finally what the responsibilities and duties of prospective parents are in reproductive medicine as compared to the rights of an unborn child (Clarkeburn 2000, Vehmas 2002). In this chapter, I will focus on a different problem, namely how assessments of 'good life' are made technically feasible during the course of selective reproductive practices. Rather than attempting to resolve very much open ethical questions – e.g. What is a life worth living? Is termination of pregnancy acceptable under any

circumstances? Who should make decisions about whether or not to terminate a pregnancy? – I will instead map out the practices that currently enable assessments of vitality, however contested these assessments may be.

To do so, I will examine ongoing attempts to stabilise and delimit the contested category of 'serious disease' in the context of selective reproductive practices in England today. In accordance with principles of informed choice and consent, it is emphasised that decisions about whether or not to begin or terminate a pregnancy must be made by prospective parents in consultation with their doctors. As Rosamund Scott (2005a, 2005b) has shown in her analysis of the cleft-palate case, the legal definition of 'seriousness' remains contested. When asked to clarify their position, the Royal College of Obstetricians and Gynaecologists has suggested that since 'there is no precise definition of "serious handicap"… the RCOG believes that the interpretation of 'serious abnormality' should be based upon individual discussion agreed between the parents and the mother's doctor' (RCOG 2008).

There have been a number of studies that have focussed on the interactions that take place in such consultations between health practitioners and patients in the context of carrier, preimplantation and prenatal testing. For example, in an analysis of health practitioners' views on 'non-directiveness', Williams et al. suggest that 'for practitioners, the boundary between choice and coercion... is not a clearcut one' (2002b: 345). Based on observations of genetic counselling sessions, Pilnick has argued that 'one of the reasons why genetic counsellors may appear to give advice or suggest courses of action in the face of the stated aim of non-directive counselling may be due to [an] ambiguity of role [since] the work of genetic counsellors may encompass anything from facilitating decision making in relation to genetic testing through to

diagnostic news delivery' (Pilnick 2002: 85). Franklin and Roberts have analyzed preimplantation genetic diagnosis patients' deliberations in terms of 'reproductive accounting' – 'how couples weigh their odds or chances in order to reach a decision about continuing treatment, and how they account for, or explain their actions' (2006: 164). And Rayna Rapp has argued that when communicating the results of amniocentesis to pregnant women, 'counselors are caught between the need to sound authoritative and the desire to "glide on the patient's wavelength"' (1988: 151).

What I will focus on instead is the burgeoning literature – in the form of pamphlets, booklets, parent guides, handbooks and websites – aimed at parents who are contemplating undergoing or have undergone carrier testing, preimplantation genetic diagnosis or prenatal diagnosis. Such information is prepared by reproductive medicine clinics, patient support groups, the National Health Service as well as disease advocacy organisations. I will also cover documents and public consultations prepared by such organisations as the Human Fertilisation and Embryology Authority (HFEA), the Royal College of Obstetricians and Gynecologists (RCOG) and the Human Genetics Commission specifically on the topic of selective reproductive practices.

By analysing these materials with a specific focus on Spinal Muscular Atrophy (SMA), Cystic Fibrosis (CF) and Down's Syndrome, I will show what concepts, norms and techniques are deployed in attempts to determine what good life is, and how these in turn are used to situate and justify selective reproductive practices. My key argument will be that while each of these three conditions have been researched and characterised in terms of their biological aetiology and pathology, what is of crucial importance in decisions about selection is how these conditions are seen to impact on a person's and/or family's 'quality of life'. Building on Ian Hacking's work around 'human kinds'

(Hacking 1995, 2002), I will suggest that not only do Spinal Muscular Atrophy, Cystic Fibrosis and Down's Syndrome entail certain biological 'modes of living' in Canguilhem's sense (Canguilhem 1989), they also entail certain 'kinds of living'. In this latter sense, life is not an anatomical, cellular or molecular affair, rather it is something that is lived, experienced, coped with, taken advantage of and improved in terms of 'quality', 'hope', 'capability' or 'happiness' (Wahlberg 2007).

**Detecting abnormality**

It is vitality that is at stake in practices of selective reproduction. If in the past, selective reproduction was about protecting and improving some kind of collective vitality (e.g. 'population stock' or 'population quality') by preventing persons of 'inferior quality' from reproducing, these days it is argued that selective reproductive practices are aimed at protecting/ensuring the individual vitality of pregnant women (as well as that of their family members) and/or her future child by allowing couples to make informed choices about whether or not to begin or terminate a pregnancy. Carrier screening, preimplantation genetic diagnosis (PGD) and prenatal screening are all elements in this process. Each involves some kind of 'non-directive counselling' where medical experts aim 'to explain the facts as clearly as possible, giving the person or family accurate information on their options in a way which they can understand' (Clinical Genetics Department 2008).

In each of these forms of selective reproductive practice – carrier testing, embryo biopsy and prenatal diagnosis – it is primarily[2] the possibility of a child being born with

[2] PGD can also be used to select embryos which have a better chance of leading to pregnancy, for sex selection or to select an embryo which would result in a so-called 'saviour sibling', and PND techniques

a disease or condition (whether hereditary or congenital) that is being addressed. Carrier testing aims at identifying 'a healthy "carrier" whose children could be affected with a particular genetic condition… [that] can cause problems', thereby affecting 'family planning decisions or other plans for the future' (Clinical Genetics Department 2008, Guy's and St Thomas's Hospital 2002). Carrier testing for conditions such as Tay Sachs Disease and Cystic Fibrosis, has been available in the UK for so-called 'at risk' (because of family history or ethnic background) populations since the 1980s. Individuals or couples identified by such testing as being substantially at risk of transmitting a disease through 'natural conception' can choose between accepting the risks, having no children, adopting, using gamete donors or using PGD and/or prenatal testing.

PGD, where a couples' gametes are fertilised in vitro and the resulting embryos are biopsied before a decision is made about which embryos to implant, is described by the Clinical Genetics Department at Guy's Hospital in London as 'a specialised treatment for couples who carry an inherited genetic defect that could cause serious health risks for their children' (Guy's and St Thomas's Hospital 2008). It is still a relatively new technique with 134 cycles of PGD carried out in the United Kingdom in 2005, resulting in 17 live births. At Guy's Hospital, one of the leading PGD centres in the UK, a total of 100 PGD babies had been born by the end of 2006. Neither carrier testing nor PGD involves medical abortion as they take place before pregnancy

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can also be used to determine the sex of a foetus which may lead some couples to terminate a pregnancy for 'social reasons' (even though abortion in the UK is not permitted on the grounds of sex alone). Indeed, it is the increasing use of selective reproductive techniques for 'non-medical' purposes that is seen by some as a dangerous slippery slope (see Duster 2003; Kerr and Shakespeare 2002). In this paper, I will focus on carrier testing, PGD and PND to prevent transmission of disease.

commences, either prior to conception or prior to implantation. Still, according to the logic of these practices, there are some inheritable conditions which some parents may wish to prevent being transmitted to future offspring.

Prenatal diagnosis using amniocentesis or chorionic villus sampling, on the other hand, can lead to the termination or continuation of a pregnancy depending on the decision of the couple. Following blood tests, ultrasound examinations, amniocentesis, chorionic villus sampling and/or, more recently, 'free foetal DNA' testing, prospective parents are given information concerning the chances that their child will be born with a certain chromosomal abnormality, hereditary disease or congenital malformation. Based on this information and following discussions with their doctor, couples will then make 'informed decisions' about whether they will continue their pregnancy or terminate it by assessing on the one hand, whether there is a substantial risk that the child will be born 'seriously handicapped', and on the other, whether they would be able to cope with caring for a child with a particular condition. As described by the NHS, prenatal testing is 'a method of detecting serious, or potentially serious, disorders in the unborn child... If a serious abnormality is detected, amniocentesis gives parents the choice of whether they want to have a child with the abnormality, or whether they would prefer the pregnancy to be terminated at an early stage' (NHS 2009).

Of the almost 198,500 legal abortions carried out in England and Wales in 2007, about 1% of them (1,939) were because a substantial risk of serious handicap was deemed (see Table 5.1). And so, despite advances in forms of genetic testing and reproductive medicine, termination of pregnancy remains the most prevalent form of selective reproduction.

**Table 5.1: No. of legal terminations according to grounds given, 2007**

| | No. |
|---|---|
| Risk of injury to the physical or mental health of the pregnant woman or any children in her family, account may be taken of the pregnant woman's actual or reasonably foreseeable environment (so-called 'social abortions') | 195,826 |
| Substantial risk that child would be "seriously handicapped" | 1,939 |
| To prevent grave permanent injury to physical or mental health of pregnant woman | 622 |
| Risk to woman's life, to save woman's life | 112 |

Source: Department of Health (2008)

Most of the different forms of testing require biological samples obtained through biopsy. Originating from a prospective parent (blood, saliva), pregnant woman (blood, amniotic fluid), an embryo (blastomere) or a foetus (free foetal DNA, chorionic villus), such samples are biochemically and genetically analysed to detect abnormalities in alpha-fetoprotein levels using blood chemistry analysis techniques, numerical and morphological chromosome abnormalities using karyotyping techniques or the presence of specific gene defects using Polymerase Chain Reaction DNA-analysis techniques. Ultrasound visualising technologies, on the other hand, allow for biometric assessment of foetuses to detect abnormalities in the amount of fluid at the back of a foetus' neck, femur length, biparietal diameter, abdominal circumference or head circumference. They also allow for morphological assessment as doctors look for 'lemon signs', 'banana signs', 'strawberry-shaped heads', 'golf balls' or other abnormal shapes which have been associated with certain conditions such as spina bifida or Edwards syndrome.

The point being that each of these diagnostic tests has been designed to detect abnormalities using genetic, chromosomal, biochemical, morphological or biometric markers associated with specific diseases or conditions. Abnormalities are detected

against 'normal' blood substance levels, gene sequences, karyotypes and foetal morphologies which have been stabilised through cumulative aggregation of clinical data. And so it is *elevated* blood substance levels, *irregular* numbers or arrangements of chromosomes, *deleted* or *mutated* gene sequences in a chromosome, *deviating* biometrics, and/or morphological *anomalies* that are singled out for further attention following diagnostic tests. Information is conveyed to prospective parents in the form of probabilities and chances as individual markers or combinations of markers are used to calculate risks. For example: if both prospective parents are identified as carriers of Cystic Fibrosis there is a 25% chance that a 'natural' pregnancy would result in a child with CF; levels of alpha-fetoprotein in a pregnant woman's blood together with her age are used to calculate chances of having a pregnancy with Down's syndrome (if the risk is calculated at more than 1 in 250, the pregnancy is classed as 'high risk'); and karyotype analysis following amniocentesis is considered to be 95-99% accurate in identifying chromosomal abnormalities in a foetus.

**Table 5.2: Breakdown of prenatally-diagnosed conditions resulting in termination, 2007**

| | **No.** |
|---|---|
| **Congenital malformations – nervous system** | **473** |
| - anencephaly | 144 |
| - spina bifida | 117 |
| - other malformations of the brain | 78 |
| - encephalocele | 33 |
| - hydrocephalus | 31 |
| - other | 70 |
| **Congenital malformations – other** | **412** |
| - musculoskeletal system | 125 |
| - cardiovascular system | 114 |
| - urinary system | 74 |
| - respiratory system | 11 |
| - other | 88 |
| **Chromosomal abnormalities** | **747** |
| - Down's syndrome | 437 |
| - Edward's syndrome | 129 |
| - Patau's syndrome | 63 |
| - other | 118 |
| **Other conditions** | **307** |
| - family history of heritable disorders | 145 |
| - fetus affected by maternal factors | 101 |
| - hydrops fetalis | 32 |
| - gestation and growth disorders | 12 |
| - other | 17 |

Source: Department of Health (2008)

Now, what is important to underscore is that while these various diagnostic tests generate biological results which give parents an idea of the chances that their child will be born with a certain disease or condition, they do not tell them anything about whether or not the disease or condition in question is serious. This involves an entirely different form of assessing vitality, and it is this form of vital assessment that will be the focus of the remainder of this chapter. In the following, I will show how the question of 'seriousness' is contested and stabilised in screening for Cystic Fibrosis carriers, PGD embryo screening to avoid transmitting Spinal Muscular Atrophy disease and prenatal screening to identify Down's syndrome pregnancies. In each case, we will see how the problem of selection (i.e. whether to begin, terminate or continue a pregnancy) is linked to estimations of 'seriousness' which in turn rely on temporal notions of onset and life expectancy on the one hand, and experiential notions of severity, suffering and quality of life on the other. Indeed, I will suggest that in reproductive medicine today, it is serious disease as certain 'kinds of living' rather than as biological abnormality, error or inferiority that informs selective practices.

**'Faulty' modes of living**

In his analysis of how different concepts of pathology have historically instigated novel understandings of biological normativity, Georges Canguilhem concludes that "there is no life whatsoever without norms of life, and the morbid state is always a certain mode of living" (Canguilhem 1989: 228). With disease come new vital norms as bodies adapt

to new conditions. "Life does not recognise reversibility"[3] and "every state of the organism, insofar as it is compatible with life [for however long], ends up being basically normal" (ibid.: 196, 200). His point being that until death silences the organs once and for all, all modes of living, even morbid ones, have their biological normality which in turn normalizes the "sick living being" in defined, if not narrowed conditions of existence. And so, however serious they might be considered, Cystic Fibrosis, Spinal Muscular Atrophy and Down's syndrome nevertheless result in certain biological modes of living for those affected by them, albeit ones which, as we will see, are judged by some to be of lower quality because of the restrictions and limitations they entail. So, how is value attached to certain modes of living over others? To answer this, let us first look at what modes of living these diseases or conditions are seen to engender.

Before any kind of diagnostic tests are carried out, prospective parents are advised to prepare themselves well by knowing their options and deciding which of these options are appropriate for them – e.g. continuing or terminating a pregnancy following prenatal diagnostic tests. It is also recommended that this preparation includes getting to 'know more about the disorder or disorders which can be detected' (RCOG 2006: 7), 'it may be important to you in the future to know that when you made your decisions, you had all the information you needed' (Antenatal Results and Choices 2007: 2). As a consequence, there is an abundance of detailed information made available to prospective parents in the form of hospital leaflets, booklets and pamphlets prepared by the National Health Service, patient groups, disease advocacy organisations not to

[3] As Sarah Franklin has pointed out, the development of Somatic Cell Nuclear Transfer or 'cloning' techniques in the 1990s has troubled this 'biological fact' as "adult body cells are induced to deliver functions they were formerly presumed to have lost", i.e. recapacitated (Franklin 2007: 32-43). The same can be said of recent development of induced Pluripotent Stem cells through genetic reprogramming.

mention a wealth of internet sites. Such information papers, parents' guides and websites have been designed to give people a concise idea of what a particular condition consists of so that they can envisage the prognosis for a child born with the condition. This includes information about the biological causes of the diseases or conditions as well as related diagnostic options.

In literature aimed at prospective parents, CF, SMA and Down's syndrome are each described as resulting from errors – mutations (3 deleted base pairs) in the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7, a missing or altered survival motor neuron (SMN1) gene on chromosome 5, and an extra chromosome 21 respectively. It is these genetic errors and resulting modes of living that are identified as the causes of the pain, discomfort and/or limitations experienced by those who are born with these conditions. With Cystic Fibrosis, parents are informed, 'the faulty gene allows too much salt and not enough water into your cells, which results in a build up of thick, sticky mucus in your body's tubes and passageways' (NHS 2008a). For a person with SMA, the missing SMN1 gene:

> makes them unable to produce Survival Motor Neuron protein. Without this protein, motor neuron cells in part of the spinal cord deteriorate and die. As a result, nerve impulses are unable to get through to the muscles that these motor neurons control, which become weaker and shrink due to lack of use. (NHS 2008b)

Down's syndrome, on the other hand, is not a single-gene disorder but rather a chromosomal disorder and it is not known exactly how an extra chromosome 21 leads to the learning difficulties, reduced muscle tone (hypotonia), flat facial profile, upward

slanting eyes as well as health problems that prospective parents are told can characterise a person with Down's syndrome (see DSA 2006b).

> Usually, the nucleus of each cell contains 23 pairs of chromosomes – 23 we inherit from our mother and 23 we inherit from our father. In people with Down's syndrome the cells contain 47 chromosomes, with an extra copy of chromosome 21. This additional genetic material results in Down's syndrome. As yet we do not know what causes the presence of an extra chromosome 21. It can come from either the mother or the father. There is no way of predicting whether a person is more or less likely to make an egg or sperm with 24 chromosomes. (DSA 2006b: 1)

In contrast, in the cases of Cystic Fibrosis and Spinal Muscular Atrophy, parents are told that they may be 'carriers' of the culpable genetic errors and therefore may be at risk of transmitting these errors to an offspring. As such, parents are advised that while carrier testing is relevant for CF and SMA, it is not applicable for Down's syndrome which at this time can only be detected after conception. Moreover, since it is almost impossible to predict which couples will conceive a child with Down's syndrome, PGD is rarely a realistic option, whereas couples where both partners are known to be carriers of CF or SMA may well opt for PGD as they are informed they have a 25% chance of giving birth to a child with that condition. Prenatal testing for Down's syndrome is offered to those prospective parents who are judged to be 'at risk' following routine antenatal ultrasound scans.

**Normal lifespan and spans of normal life**

As I have already suggested, conditions like CF, SMA or Down's syndrome do not only denote certain 'faulty' modes of living, they also denote particular 'kinds of living'. This becomes clear in the descriptions given to prospective parents of the symptoms and limitations *experienced* by those individuals who have been born with CF, SMA or Down's syndrome, i.e. the impact they have on individuals' lives. With Cystic Fibrosis, the National Health Service informs parents that 'many parts of the body are affected including the pancreas and its secretions, which leads to malabsorption, malnutrition and vitamin E deficiency, and the lungs, which results in frequent chest infections and lung damage... Median life expectancy for patients with CF is around 31 years' (NHS 2008a). And, in their information materials, the Cystic Fibrosis Trust describes it as 'the UK's most common life-threatening inherited disease… affect[ing] over 8,000 people', again highlighting that 'average life expectancy is around 31 years, although improvements in treatments mean a baby born today could expect to live for longer' (Cystic Fibrosis Trust 2000).

**Table 5.3: Types of SMA**

| | |
|---|---|
| **Type 1 (Severe)** | Also known as Werdnig-Hoffman Syndrome. Onset before or shortly after birth. Unable to sit. Do not usually survive past 2 years old. |
| **Type II (Intermediate)** | Onset between 3 months and 2 years. Able to sit, but not stand without aid. Survival into adulthood possible. |
| **Type III (Mild)** | Also known as Kugelberg-Welander Disease. Onset usually around 2 years. Able to walk. Normal lifespan. |
| **Adult Onset SMA** | Number of forms differing in age of onset. Degree of weakness is variable. |

Source: Jennifer Trust (2008a)

Spinal Muscular Atrophy is described by the Jennifer Trust for SMA as affecting 1 in 6,500 babies born (about 260 per year) and as the 'biggest genetic killer of infants

in the UK' (Jennifer Trust 2008b). As a condition it is divided into 4 types which have been graded by the Trust in terms of severity, with life expectancy being one of the key indicators of this severity (see Table 5.3).

While stressing that Down's Syndrome is 'not a disease', the Down's Syndrome Association (the UK's largest support group for people living with Down's Syndrome) describes it as one of the most common genetic conditions affecting around 60,000 people in the UK. In 2006, 1,877 diagnoses of Down's syndrome were made in England and Wales of which 1,132 (60%) were prenatally diagnosed. There were an estimated 749 live births, although following confirmed prenatal diagnosis an estimated 1,000+ pregnancies were terminated with 436 of these terminations primarily justified on the grounds of a substantial risk that if born the child would be 'seriously handicapped' (Department of Health and National Statistics 2007, NDSCR 2006).[4] Nevertheless, although there are clearly many prospective parents who do view Down's syndrome as serious enough to warrant a termination of pregnancy, information available to prospective parents considering their options inform them of dramatic improvements in a Down's syndrome child's prognosis over the last half century or so:

> In the 1950s, many people with Down's syndrome did not live past the age of 15. However, due to a better understanding of the condition, and

[4] While there are no available statistics on this, presumably the other 500+ terminations were carried out on the legal grounds that a medical termination of pregnancy was necessary to protect the pregnant woman's physical and mental health (Scott 2005b: 310). Also, a recent survey by the Down's Syndrome Associations showed that in 2006 for the first time there were more live births (749) than in 1989 (717) when screening became available, a 15% rise after taking into account the UK's overall rise in birth rates (BBC News 2008).

> advancements in treatment and care, the average life expectancy of someone with Down's syndrome is now 60-65 years of age. (NHS Choices 2008)

Survival then, is one factor used to gauge seriousness. Yet, while a 'normal lifespan' is one of the norms against which seriousness is measured, there is no clear cut off point but rather continuums. The genetic errors behind CF, SMA and Down's certainly can impact catastrophically on bodily vitality and thereby shorten a person's lifespan (compared to average life expectancy) even to under two years in the case of severe SMA, yet others can live to 30 or even over 60 years.

Another temporal component in determinations of seriousness is that of onset, and again we can see that there is considerable variability with some conditions manifest at birth, some manifesting themselves 'around 2 years' and others much later in adulthood. Indeed, in 2006, the HFEA for the first time approved embryo testing for susceptibility genes associated with inherited cancer pointing out that 'these conditions differ from those already licensed before because people at risk do not always develop cancer, it may occur later in life and some treatments may be available' (HFEA 2006). So, in some cases, the mere possibility that a condition will set in at a later point (perhaps never) is deemed 'to be sufficiently serious to merit the use of PGD embryo testing' (ibid.) so as to avoid implanting susceptibility-gene-affected embryos, or put in another way, to prevent potentially faulty modes of living from coming to term.

With onset, it is not so much a 'normal lifespan' as a span of 'normal life' (prior to onset) that is the norm against which seriousness is measured, and again, onset

appears as a continuum with no clear cut off point.[5] In terms of selective reproductive practices, some of the targeted serious diseases are congenital (present from birth) while others may appear in an affected individual much later in life (if ever). It is noteworthy that in their report on new reproductive technologies the Human Genetic Commission suggested that 'a distinction may be drawn between the moral status of an unimplanted embryo and a fetus in an established pregnancy, and that this distinction may be used to justify the use of PGD for certain conditions where prenatal diagnosis cannot be regarded as appropriate or acceptable' (Human Genetics Commission 2006: 49). One such case was testing for serious late onset conditions.

Normal lifespan and span of normal life are social rather than biological norms which, in a sense, organise disease variability in terms of vital continuums that are used to prognose a patient's vitality over time. The former allows for assessment of a disease in terms of its impact on the length of an affected person's life – by how much is a disease expected to shorten a person's life when compared against some kind of (usually national) average life expectancy? The latter, on the other hand, provides an indication of how much 'normal life' an individual can expect to live before he or she is affected by a disease that lies dormant in his/her genes. Upholding both norms is some kind of notion of a 'normal life' as opposed to an affected life. That is to say, CF, SMA and Down's syndrome are diseases or conditions which can be accounted for not only in terms of faulty gene expression or regulation, but also as certain 'kinds of living'. With

[5] Barbara Katz Rothman (1998: 186) captured this gradated fluidity in her reflections on 'spoiled pregnancies': "Anencephalic babies live for a few days. Tay-Sachs babies live for a few years. Children with cystic fibrosis lived a decade or two, longer now with better treatment; some of the familial cancers come in people's 30s; Huntington's disease comes at midlife. There is no point, we say, in continuing the pregnancy if the baby is going to die right away. How about soon? How soon?"

this term I borrow from Hacking's notion of a 'human kind' which he argues differs from a 'natural kind' because 'the classification of people and their acts can influence people and what they do directly' (Hacking 1992: 190). Diagnosis is a prevalent act of classification and although the genetic errors that are seen to cause CF, SMA and Down's syndrome as modes of living are a necessary diagnostic criteria, it is social rather than biological norms that are invoked when ascertaining the 'severity' or 'seriousness' of a particular disease or condition. As kinds of living, diseases or conditions can be deemed inferior both in terms of shortened lifespan and/or in terms of a shortened span of 'normal life' for the affected individual. Value is attached to living long (as related to average life expectancy), unaffected (compared against 'faulty' modes of living) lives.

**Living with…**

But it is not only temporal norms of vitality that are relevant in deliberations about the seriousness of a disease or condition. Perhaps more important is the concept of 'quality of life' and the vital norms it entails, as further to the clinical descriptions of symptoms, onset and life expectancy discussed above, prospective parents are also provided with considerable amounts of qualitative information based on interviews with people who have been diagnosed with a certain disease as well as their parents and siblings. These booklets and websites have titles like 'Living with Cystic Fibrosis', 'Down's syndrome – a new parents guide', 'Personal stories – Type 1 SMA' or 'Cystic Fibrosis and You'. In these accounts, it is not so much genetic errors, medical symptoms and life expectancies that inform prognoses, rather it is patient experiences, coping strategies and condition management advice. Whatever the limitations imposed on a child by

these conditions (learning difficulties, immobility, tiredness, pain, poor immunity, etc.), it is argued that they can nevertheless be fulfilling according to their own terms.

For example, the UK's Down's Syndrome Association argues that if individuals with Down's syndrome have had a low quality of life this has more to do with prejudice than with their condition:

> In the past it was believed that there were many things that people with Down's syndrome could not do when in fact they had never been given the opportunity to try. Today these opportunities have never been greater with many people with Down's syndrome leading rich and varied lives. People with Down's syndrome are now leaving home, forming relationships, gaining employment and leading independent and active lives with differing levels of support. The quality of life, life expectancy and role in the community for adults with Down's syndrome has been transformed as education, support and opportunities have improved. (DSA 2007: 12)

With CF, it is stressed that '50% of people with CF now live into their late 30s but the condition can severely affect their quality of life' (CGE 2007: 3-4) making it 'vitally important that those with Cystic Fibrosis receive appropriate healthcare to ensure a better quality and length of life' (Cystic Fibrosis Trust 2008a). At the same time, it is also highlighted that most children with CF will just want to go on leading 'a normal life' at school, with friends, etc. – 'I just want to live a normal life really, just get on with it. Brave is a horrible word, never use it' (Youth Health Talk 2008).

'Even' in cases of SMA Type 1 where life expectancy is rarely above 2 years of age and the child suffers from poor cough, poor feeding and chest infections, information for parents includes 'Tips to Improve Quality of Life' such as providing the

child with 'Light and Sound toys of all types to stimulate your child's imagination' (Jennifer Trust 2008d). A study from 2003 which compared healthcare professionals' assessment of quality of life of children with SMA Type 1 with that of their primary carers concluded that 'although there is a widespread perception that spinal muscular atrophy type 1 children have a poor quality of life, this perception is not shared by their care providers' (Bach, et al. 2003: 137).

What is more, in patient literature about each of the three conditions it is consistently pointed out that 'individuals [with SMA] vary enormously' (Jennifer Trust 2008a), 'there is no such thing as a typical person with Down's syndrome… some have more serious difficulties than others' (NHS 2006: 3) or that 'CF affects people in a lot of different ways – some have it severely, but many have it mildly or moderately' (Cystic Fibrosis Trust 2000: 7). The way in which each condition affects individuals can be graded according to severity understood as a kind of intensity of symptoms, limitations or suffering.

Notwithstanding these abundantly available 'living with' accounts of certain diseases or conditions which maintain that all modes of living (however 'faulty'[6]) have

[6] The question of whether or not there are any kinds of living that are so poor as to be not worth living has been discussed by many (see Brody 2002; Reuter 2007). Some argue that 'only the most devastating diseases, such as Tay-Sachs disease or Lesch-Nyhan disease… involve so much pain and suffering before death with so little in compensating benefits that those suffering from them are properly described as having lives not worth living' (Brody 2002: 347), while others maintain that '*every life* is worthy of our protection, our care, and our welcome. No one should ever discount the difficulties of dealing with children who are born with severe genetic abnormalities or serious diseases… Nevertheless, these are the very same issues we will all face in terms of issues at the end of life, and at many points between birth and death' (Mohler 2006, emphasis original). In a parent's guide, one couple describes the moment they

some kind of quality, normative estimations about the quality of life of persons affected by CF, SMA or Down's are a key component in deliberations about whether to begin, terminate or continue a pregnancy. The Cystic Fibrosis Trust, for example, which helps families and individuals who suffer from CF underscores that 'while there have been great improvements in the length and quality of life for people with CF, it still remains a serious condition and carrier couples should think very seriously before undertaking a pregnancy' (Cystic Fibrosis Trust 2002: 7). And a father of a child who died from SMA writes on the Jennifer Trust's website:

> Why use PGD? Because it works! Individual families known to be carriers of genetic disorders are always faced with difficult decisions when contemplating the start or the continuation of family life… Saying that it would be better for a child to be free of the disease does not necessarily reflect on attitudes towards those with the disease… For anyone who knows what it is like to care for a severely disabled child, the difference between having a child with and without a condition is not one of love and care of the child but about the impact that the extreme disability has on the family and affected child. (Jennifer Trust 2008e)

When the HFEA approved PGD for certain late onset diseases they made a point of stressing that:

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decided not to have their son who had Tay Sachs disease resuscitated: 'one day when he was about 2½ and had to be admitted to the hospital. He was very weak. He was having trouble eating and drinking. We knew at this point it was not possible for him to have any quality of life, and it was at this point we made that decision. It was very difficult to make, and prior to his getting this sick I would not have been able to make that decision.' (Borfitz and Margolis 2006: 11).

> The role of medicine has always been to try to relieve pain and suffering and to try to improve the quality of life for people... [Our] decision today deals only with serious genetic conditions that we have a single gene test for. We would not consider mild conditions – like asthma and eczema – which can be well-managed in medical practice… The Authority agreed that we should consider the use of PGD embryo testing for conditions such as inherited breast, ovarian and bowel cancers given the aggressive nature of the cancers, the impact of treatment and the extreme anxiety that carriers of the gene can experience. (HFEA 2006)

With so many variables in play (severity, suffering, pain, dependency, immobility, disability), it is little wonder that there is no consensus when it comes to determining which diseases are serious using quality of life criteria. The UK's Human Genetics Commission has summed up such variability as follows:

> it has proved difficult to define what is meant by 'serious'. One way of doing this would be to draw up a list of conditions that are considered to lead to a very poor quality of life… However, this approach fails to recognise that quality of life judgements are subjective, and that genetic disorders are variable in terms of severity and health outcomes. There is evidence to suggest that people with genetic disorders, their families and professionals all have different views about which conditions give rise to a poor quality of life. In general, those who have direct experience of *living with a genetic disorder* are likely to rate the quality of their lives more highly than would medically qualified professionals. (Human Genetics Commission 2006: 36, emphasis added)

Similarly, in connection with recent parliamentary debates to amend the Human Fertilisation and Embryology Bill in early 2008, the Royal College of Obstetricians and Gynaecologists argued that:

> a strict definition [of what constitutes a serious abnormality] is impractical because we do not have sufficiently advanced diagnostic techniques to detect malformations accurately all of the time and it is not always possible to predict the 'seriousness' of the outcome (in terms of the long-term physical, intellectual or social disability on the child and the effects on the family). The RCOG believes that the interpretation of 'serious abnormality' should be based upon individual discussion agreed between the parents and the mother's doctor. (RCOG 2008)

And so it is in practice; agreement on termination of pregnancy on the grounds of substantial risk that the child, if born, would be seriously handicapped, comes about through consultation between prospective parents and their doctors, with two doctors having to authorise the termination by signing a so-called Certificate A form. The point being that determination of 'seriousness' has less to do with biochemical or genetic test results from the laboratory and much more to do with discussion and information exchange between prospective parents and doctors about what living with a certain disease or disorder entails. To reach agreement on whether a disease or condition is serious, prospective parents are not given blood substance level values or karyotype maps, rather they are provided with qualitative information which presents other patients' and families' experiences of 'living with a genetic condition'.

Once again, vital norms emerge in attempts to organise variability of 'interpretation' along continuums of quality of life, where value is attached to living

independently, having social relationships, gaining employment and of course not suffering from the symptoms caused by genetic conditions. As kinds of living, CF, SMA and Down's syndrome have their own particular vital norms which make space for, albeit narrowed, continuums of not just lifespan and onset, but also quality of life particular and relevant to these conditions. Borrowing from Canguilhem, we might say there is no living without norms of living, and living with a genetic disease or condition is always a certain kind of living. Yet, at the same time, these kinds of living are also assessed against the norms of living associated with those who lead a 'normal life'. This is the case when some prospective parents decide that there are some kinds of living which should be prevented from coming into being for the sake of the child; that some 'faulty' modes of living are not worth living. And so, where some might consider Down's syndrome serious enough to warrant termination of pregnancy because of the condition's potential impact on the future child's quality of life, others would vigorously dispute this by arguing that people with Down's syndrome can 'live full and rewarding lives' (DSA 2008). Such contradictions are not resolvable by recourse to biology.

**Coping**

When it comes to living with a genetic condition it is of course not only the affected individual who is living with the condition. In a very concrete and intimate sense, so too are the parents and siblings of the affected individual as it is they who will, in by far most cases, be caring for the affected child from birth and often throughout his or her life. We have already seen how, in cases where parents are considering termination of a pregnancy, negotiation of what constitutes seriousness currently takes place in

consultation between prospective parents and their doctors. To carry out PGD in the UK, on the other hand, clinics must be licensed to do so and may only offer PGD for diseases and conditions that have been approved by the Human Fertilisation and Embryology Authority. In both cases, it is not only the impact of the condition on the child (if born) that is taken into consideration, it is also 'the effects on the family' (RCOG 2008) or 'the way it affects the family' (HFEA 1999). Moreover, as we saw earlier, the decision to license some late onset disorders for PGD was partly based on 'the extreme anxiety that carriers of the gene can experience' (HFEA 2006). As put by the Human Genetics Commission, '[reproductive] decisions are often linked to whether the family feels it could cope with the demands of a child with such problems, the impact it would have on other children, or on the carers' (2006: 3).

As such, literature provided to prospective parents also includes information on the difficulties that other parents have had in coping with caring for a child with a genetic disorder. Prospective parents are also encouraged to get in touch with other parents who have given birth to and cared for a child with a similar genetic condition. Through such interaction, parents are reassured that they are 'not alone' as they learn how 'most parents find out that their baby has Down's syndrome soon after the birth and the news is a great shock', how parents can be left 'feeling confused, angry, alone or afraid' after a diagnosis of SMA or how 'coping with CF at the time of diagnosis… can be challenging' (Cystic Fibrosis Trust 2008b, DSA 2007, Jennifer Trust 2008b).

Carers' descriptions of living with a genetic condition in terms of coping are of course not limited to the moment of diagnosis, for once a diagnosis is confirmed parents will often want to do everything to ensure that their affected child is given the best possible life under the circumstances. This can be a challenging task to say the least:

Parents' ways of coping with their children with CF differ as widely as the condition of the children themselves. The whole family – the parents, the child or children with CF, other siblings – will all be affected by the psychological pressures arising from the chronic nature of CF, the uncertainty about the future, the genetic aspects, worry, depression and the tiring routines of physio and supervising medication. (Cystic Fibrosis Trust 2008b: 9)

The demands of living with a young child can be overwhelming particularly when the fact that your child has Down's syndrome may lead to extra appointments with doctors and therapists and anxiety in the early years. (DSA 2007: 11-12)

At the back of our minds we did keep alive the possibility that she might not have Down's syndrome but we knew that we would be able to cope if she did – there's so much out there for her. Schools are integrated and there are even actors with Down's syndrome. There's a worker at our local supermarket who has Down's syndrome and we think that it doesn't need to hold you back. (BBC News 2008)

We were determined that Amar [diagnosed with SMA Type 1] would have the best time we could imagine... We never complained about the sleepless nights, possibly three hours of sleep a night on average. My wife stayed at home with Amar all day, every day, until I came home from work in the evening. He stayed with me, so I could give my wife a break. We were committed and got used to it, and enjoyed it, even though it was hard. We never complained. We coughed every cough for him; we wanted him to stay as well as possible. (Jennifer Trust 2008c)

The argument that the impact of a genetic condition on a family's life is relevant when determining what constitutes seriousness is perhaps one of the most controversial. As Scott has shown, in a 'wrongful birth' case from 2000 in East Dorset which was brought by parents who argued that they had lost the opportunity to abort a child with Down's syndrome because of a breach of duty, the presiding judge concluded that 'the birth of a disabled child will dramatically affect the quality of life of both parents and it is to be inferred that a reason why they would have terminated the pregnancy was to avoid such a loss of amenity in their lives' (cited in Scott 2005a: 402). A distinction is made between caring for a 'normal child' and one affected by a condition such as Down's syndrome ('She will need care and supervision for the rest of her life' (ibid.: 401)). What is more, it is a normative distinction since looking after an affected child is seen to negatively impact the quality of life of the couple.

Such arguments have been controversial especially because they are considered to discount the interests of the future child in favour of the 'selfish' interests of the parents. It is also suggested that once the future child's interests are set aside, the problem of 'serious disease' can very quickly become framed in terms of burden, whether this burden is considered psychological, social, emotional, financial or genetic. For example, Tom Shakespeare has argued that if sufficient care is not taken 'decisions about reproductive choices are likely to be influenced by the fact that an unjust society means that having a disabled child places a severe financial and practical burden on a family' (Shakespeare 1998: 679). And if reproductive selection becomes a matter of alleviating burdens for collectives (e.g. 'family', 'society', 'population', 'human gene pool') then

reproductive medicine is once again well on its way down the slippery slope to eugenics (see also Wahlberg 2008).

Notwithstanding these controversies, it is clear that the circumscription of genetic conditions as 'kinds of living' takes place not just as regards the affected individual but also his or her carers. The perceived impact a genetic disease or condition has on a family's quality of life is a central element in deliberations about what constitutes a serious disease. Indeed, in the case of the termination of a pregnancy in week 28 following prenatal diagnosis of a cleft palate with which I started this chapter, the curate argued that it was an 'error of law' that the medical practitioners who authorised the termination 'took into account the views of the parents involved' (cited in Scott 2005b: 309). In contrast, as we saw earlier, the RCOG suggests that interpretation of what is meant by 'seriously handicapped' should be resolved between parents and doctors.[7]

The birth of a child affected by a genetic condition also introduces a new 'kind of living' for carers and siblings as their lives are transformed. Since there is no living without norms of living, it follows that this new kind of living for families will have its own norms, however 'narrowed'. And, just as there is no consensus concerning how good a quality of life persons affected by genetic conditions can have, there is no consensus on whether a family's quality of life will necessarily deteriorate as a result of caring for a person with a genetic condition. Indeed, in many of the parental accounts of living with a genetic condition, it is often pointed out how such an experience can in

[7] Williams et al. (2002a: 65) have also shown how some practitioners find it hard to 'draw the line' with one of their respondents arguing: 'I mean, I've seen a woman who had a cleft lip and palate herself, her first child had a cleft lip and palate, she had another baby with a cleft lip and palate and she said, "I want a termination". Now who am I to say to her that I know more about cleft lip and palate than she does?'

fact enrich and strengthen family life – ‘I can only describe how much joy my son [who has Down’s syndrome] has given me’ (DSA 2006a: 7) .

**Conclusion – modes and kinds of living**

In this chapter, I have investigated how the estimations of vitality that inform attempts to circumscribe and stabilise ‘serious disease’ as a legal and medical category are made feasible and practicable. While the diagnosis of a certain disease or condition in reproductive medicine relies on biochemical and genetic analyses of biological samples and/or biometric and morphological analyses of ultrasound scans, there is nothing in the laboratory techniques of Polymerase Chain Reaction analysis, blood substance level analysis, karyotyping or sonography that can qualify a disease or condition as ‘serious’, ‘intermediate’ or ‘mild’.

Instead, I have shown how value is formed in the transmogrification or looping of certain diagnosed ‘faulty’ modes of living into kinds of living. As modes of living, hereditary and congenital conditions are characterised in terms of genetic errors which are ultimately seen as narrowing, to varying degrees, an individual’s biological conditions of existence. As kinds of living, these same conditions are characterised in terms of their constraining/negative impact on ‘normal life’ or ‘quality of life’ – immobility, dependence, learning difficulties, premature death, poor immunity, pain, suffering, etc. It is in this circumscription of genetic conditions as kinds of living that phenomenological nosologies emerge, making it possible, for example, to classify SMA Type 1 as ‘severe’, SMA Type 2 as ‘intermediate’ and SMA Type 3 as ‘mild’, to suggest that ‘some have CF severely, but many have it mildly or moderately’, or to classify breast and bowel cancer as ‘serious’ and asthma and eczema as ‘mild’. As, we

have seen, such attempts to grade the seriousness or severity of certain diseases or conditions are often described as subjective and therefore inconsistent. Such variability and inconsistency has nevertheless not prevented ongoing efforts to do exactly this, especially when decisions about whether or not to begin or terminate a pregnancy must comply with law.

What I have argued is that when attempts are made to classify conditions or diseases according to gradations of seriousness or severity, this has been made possible by norms of living which have emerged out of qualitative interviews with individuals affected by these conditions, their parents and families as well as medical doctors. Such qualitative or human technologies – 'technologies that take modes of being human as their object' (Rose 1996: 26) – are not somehow abstract or 'merely' subjective when compared to the laboratory technologies used to diagnose certain conditions, rather they are concrete and palpable, resulting in parent guides, ranking of different Types of SMA according to severity, coping strategy manuals, guidelines for termination of pregnancy, etc. It is the social norms of living ('normal lifespan', 'normal (quality of) life', 'normal family life') that these human technologies have generated which in turn allow for assessments of a certain condition's likely impact on the life of a child and her/his family. At the same time, they have also generated new norms of living that are particular to the limitations and restrictions attributed to a disease or condition on an individual and his/her family. These condition-specific norms of living are what allow parents to nevertheless do everything they can to give the best possible lives to their affected children. There are no clear cut off points, rather there are continuums of lifespan and quality of life which are used as navigation aids in deliberations over

whether or not to begin, terminate or continue pregnancies that may result in a child being born with a serious disease or condition.

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