REFLECTIONS ON THE LAW AND ETHICS OF REGULATING PREIMPLANTATION GENETIC DIAGNOSIS IN THE UNITED KINGDOM

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Abstract

The purpose of this thesis is to query the legitimacy of offering preimplantation genetic diagnostic (PGD) testing against Down's syndrome on the basis of United Kingdom (UK) law and policies. I will argue that extending PGD testing for Down’s syndrome as a permissible use of this technology does not (straightforwardly) adhere with the Human Fertilisation and Embryology Authority (HFEA) Code of Practice's stated factors which are to be considered when assessing the appropriateness of PGD applications. Indeed, due consideration of the evidence given in the relevant literature about the capacities and quality of life possible for persons living with Down's syndrome would seriously call into question the validity of a positive judgment recommending PGD as a treatment service for Down's syndrome according to the current UK regulatory instruments. I end the thesis by considering why the HFEA's relatively recent decision to limit client access according to an exclusive list of "serious" and therefore "in principle" test-worthy genetic conditions—understood as legitimate applications for PGD—stands to entrench prejudice, stigma, social bias, and unfair discrimination against the disadvantaged social group of persons living with Down's syndrome.

Keywords

Bioethics, Disabilities, Discrimination, Down's syndrome, Ethics, Feminism, Law, Preimplantation genetic diagnosis, Regulation

Abbreviations

ARTs = Assistive Reproductive Technologies
HFEA = Human Fertilisation & Embryology Authority
IVF = In Vitro Fertilisation
PGD = Preimplantation Genetic Diagnosis
PND = Prenatal Diagnosis
UK = United Kingdom

Introduction[i]

Inherited genetic diseases are identifiable in 1-3% of the population at birth.[1:354;2] As available medical treatment for many of these diseases is still limited, current medical research in relatively wealthy, technologically-advanced societies has for (at least) the past two decades applied itself to preventing the birth of affected children.[2;3] Preimplantation genetic diagnosis (PGD) is a technique involving the biopsy and genetic testing of *ex utero* embryos created through *in vitro* fertilization (IVF). The resulting information is used by parents and clinicians to make decisions concerning which embryos to transfer for implantation to the prospective mother's uterus in the hopes of initiating a pregnancy. Pioneered for use in humans at the end of the 1980s,[4:941;5] PGD was originally developed as an alternative to post-conception diagnostic procedures[ii] for would-be parents seeking to have a genetically-related child free of, or less likely to develop, an undesired heritable genetic condition for which at least one of the parents would be a known carrier.[6]

The identification and avoidance of disease susceptibility in embryos is the most common goal of PGD.[iii;8:8] Initially PGD was used to identify and screen for the relatively small number of embryos at high risk for fully penetrant[iv] and fatal, or nearly fatal, seriously debilitating, childhood onset conditions.[6;9] This, however, has been changing as the use of PGD has been expanding. The European Society for Human Reproduction & Embryology[v] claim that PGD tests have increased from 131 IVF cycles in 1999 to 2,984 in 2003/4[11] to 6,561 PGD/PGS cycles in 2010.[12] There are now more than 2000 conditions for which genetic testing is available for use

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[i] This thesis, as per prior approval from the Graduate Studies Committee of the Departement de Philosophie, Université d'Ottawa utilizes previously written papers (now published articles) as a basis for chapters, including: Krahn, T. (2007). Where are we going with preimplantation genetic diagnosis? *CMAJ*, 176(10):1445-1446; Krahn, T. (2009). Preimplantation genetic diagnosis: does age of onset matter (anymore)? *Med.Health Care Philos.*, 12(2):187-202; and Krahn, TM. (2011). Regulating preimplantation genetic diagnosis: the case of Down's syndrome. *Med Law Rev*, 19(2):157-191.

[ii] This is often referred to as "Prenatal diagnosis" or PND. Examples include amniocentesis and chorionic villus sampling.

[iii] As a matter of practice, prospective PGD clients in the UK only present for clinical services based upon a medical practitioner's recommendation in the UK. PGD is to be distinguished from preimplantation genetic screening (PGS). PGS is not used to look for a specific disease. PGS refers to a special application of PGD where embryos from "presumed chromosomally normal genetic parents are screened for aneuploidy."[7] PGS is most commonly used as an adjunct service for clients who come to the clinic to obtain IVF services first and foremost to overcome issues related to their infertility. This thesis does not deal with PGS insofar as this involves PGD applied as a screening measure *for populations*. Instead the analysis is reserved to cases where there is a known, heritable genetic condition for a given client.

[iv] Probability that genotype will be expressed as a genetic disorder.

[v] This society has organized a data collection system of informal reporting covering about two-thirds of all PGD activity in Europe.[10]

in clinical settings,[13] and from a scientific standpoint, virtually any postnatal genetic test can be used to test embryos.[14:32] With these developing technological powers, on some viewpoints, we have the possibility of what is understood as a truly preventive medicine[vi;15:242] as well as greater control in "choosing our children"[17]—or, more accurately, what kinds of children parents do not want to have, or do not want to have again.[9;18-20]

The expansive potential for this form of genetic testing has been greeted with both enthusiasm and concern.[vii] As with other new technologies, so also with PGD, there is an accompanying public uneasiness, "especially when it is felt that scientific and medical advances are running ahead of public debate of the social and ethical issues involved."[6:1] It is a commonplace in the bioethics literature, and especially media reporting of PGD, to signal the relevant moral dangers attendant to new applications of this technology by referring to threshold metaphors, most notably that of the "slippery slope".[see 23] Tied to this casting of what is morally at stake with PGD is the view that relaxing the rules of who gets access to PGD will increase the demand for this technique to the point that it will eventually be on offer for trivial reasons, thus making possible "designer babies"[24] and a society that degrades the parental role, viewing children as mere consumer objects.[25]

Public consultations in the United States,[26;27] the United Kingdom (UK),[28;29] and Canada[30;31] have demonstrated widespread support for the use of PGD to help prevent the birth of "seriously impaired" offspring and widespread disapproval of utilizing PGD in support of mere individual or social preferences for or against non-health related traits (e.g. eye colour, sex selection for "non-medical" or "social purposes").[viii;44] The International Bioethics Committee (which includes UK representation) has recommended that "PGD be limited to medical indications" and contrasted this with the unethical practice of using PGD for "non-medical

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[vi] King has argued that "preimplantation diagnosis thus represents a new intensification of the medical surveillance of human reproduction, which would institute a far more preventive regime than currently operates for prenatal testing."[15:244, note 492] See King (1999:180).[16]

[vii] Bennett explains the complexity of "overlapping concerns about the discarding of embryos as well as concerns over the potential eugenic implications of the technology and the possibility that the technology may be used for 'non-medical' reasons."[21:170] On fears of PGD being used for eugenic outcomes, see HGC (2001:para.6).[22]

[viii] Meister et al. note that "a consensus can be seen in the fact that non-medical reasons do not legitimate the practice of this testing..."[32:232] Bredenoord et al. have noted that "[t]he objections to PGD mainly regard risk and safety issues, the discarding of embryos, the selection of embryos, the slippery slope and fears for eugenics (King, 1999).[16] Notwithstanding these objections, there is a strong consensus that PGD is justifiable as well, at least for severe genetic disorders (Fasouliotis and Schenker, 1998[33]; Buchanan et al., 2000[34]; Verlinsky and Kuliev, 2000[35]; Steinbock, 2002[36]; Robertson, 2003[37]; Klipstein, 2005[19]; Kuliev and Verlinksy, 2005a[38],b[39]; MacMahan, 2005[40]; Dresser, 2006[41])."[42:87] See also Gunning (2008).[43:35-36]

reasons".[45:15] While in the United States PGD[ix] is mostly unregulated except for standards of clinical safety (and sometimes efficacy),[41] European countries have favoured oversight in the form of both clinical and ethical regulations. As such, most jurisdictions that accept but regulate the availability of PGD restrict it to what are characterized as "serious" conditions.[48:201;49-51]

In the past decade, assistive reproductive technology (ART) professionals as well as relevant regulating bodies have been under significant pressure from a variety of sectors to expand the category of "serious" (or what are considered "serious-*enough*") conditions to facilitate better access for PGD clients.[52:para.29-30;53;54:183-186;55:6] The effect has been continuously expanding licensing practices of PGD, concomitant with continuously expanding lists of what are considered "serious" genetic conditions. These developments have met with certain public and political concern expressed as a need to draw boundaries to control the use(s) of PGD.[56-58] Indeed, reporting in the media,[x] professional journals,[60] and government reports and policy documents[23;cf.61] have generally-speaking emphasized that for moral safeguarding against ethical abuses of the technology what is needed are rules for delimiting client access.

Ethical concerns with PGD regulation

Client access is an important, though not the only, stratum for the regulation of ARTs in general, and PGD in particular. There are within the bioethics and legal academic literatures a variety of potential avenues for critiquing any approach to regulating PGD. On the one hand, there are important questions to investigate for determining who should, or should not, be made responsible for regulating PGD. What sort of authority should underwrite PGD regulators? What limits should apply to the reach and powers of any delegated authority? And how should these parameters be decided? On the other hand, there are a variety of interests implicated in the very research, development, and clinical delivery of this technology. As such, there are important questions to ask concerning, if and how the state should have a role in controlling the evolution of these technologies? What safety and efficacy standards should be applied to clinics that offer PGD? What mechanisms of clinical oversight are warranted, and who should design and who should enforce these clinical standards?

There are many vested interests at stake with PGD, with potential harms and benefits to various parties that may be difficult to harmonize, sometimes leading to social tension or even

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[ix] As with most other assisted human reproductive technology services in the United States.[46;47:ch.2,sec.III]

[x] This moral alarm is evidenced by Brownlee's reporting in the *Washington Monthly* as follows: "The nation is already sliding down a slippery slope toward the age of reprogenetics. Our only hope of slowing the pace is to apply the brakes of regulation."[59]

public conflict. For instance, much has been written in the bioethics literature on the moral status and welfare of human embryos, including those embryos that are created through IVF and then tested with PGD. Here (as we shall see in chapter 1) there are questions of whether human embryos are deserving of special protections through regulation against the sorts of harm that may result from handling, storing, testing, and sometimes even destroying IVF embryos. Some have asked whether regulation should be used to either promote or prohibit the development of certain embryos based on information acquired through PGD and what kind of changes this might or might not bring for society. Others have been concerned with questions regarding what information about embryos parents should be allowed access to and whether parental powers should be limited: for example, limits that would prevent parents from over-determining the genetic make-up of their future children.[62] Some women's groups and feminists have looked suspiciously at regulation that would impede client access to PGD as yet another form of women being forced to concede ultimate control over their reproductive projects. Other women's groups and feminists see PGD as not only a tool with which to potentially augment women's reproductive liberty but also as one that could compromise their reproductive autonomy. On their view, PGD could feed into societal expectations that women make use of these technologies or else bear responsibility for "choosing" not to prevent the birth of children with heritable genetic impairments. In this regard, some feminists and women's groups have emphasized the importance of guarding women's reproductive interests and autonomy by regulating how PGD is delivered to women in the marketplace of reproductive medicine.

Scope, focus, and aim of thesis

This criss-cross of questions and concerns is given here in order to emphasize the point that what is ethically required for regulating PGD is a multi-layered—indeed, tangled—moral space. Though many of the relevant threads of critique are inter-related, this thesis will focus specifically on disability interests and concerns that are implicated in the way that PGD is regulated in the UK. Historically, and still today, the UK approach to regulation has been to try to check the negative potential of PGD by limiting consumer access through a licensing system entrenched in law and administered by an independent regulator, the Human Fertilisation & Embryology (HFEA)—the longest, most fully developed, and many would agree, the most influential legal regime that regulates the handling and use of human gametes and embryos. In short,

> the HFEA must agree that a particular genetic condition is sufficiently serious before clinics are permitted to test for that condition using PGD. [And] even if the HFEA approves the genetic condition for testing, clinics must make their own judgment on whether PGD is the appropriate treatment for a particular patient.[63]

Down's syndrome is officially recognized by the HFEA as one of a list of (at the time of writing) 256 sufficiently serious conditions deemed "acceptable for clinics to use PGD to test for."[64] Given that some ART clinicians have used PGD as a "treatment" service to prevent implanting embryos with Trisomy 21 in the context of IVF,[65-67] the question I want to consider in this thesis is what makes it appropriate for the HFEA to authorize, medical professionals to offer, and society to allow the use of this technology for this purpose? What makes Down's syndrome "sufficiently" serious such that PGD testing for this condition is considered legally and ethically permissible in the UK?

The purpose of this thesis, then, is to query the legitimacy of offering PGD testing against Down's syndrome, not in and of itself, but rather on the basis of UK law and policies. I will make a case that extending PGD testing for Down’s syndrome as a permissible use of this technology arguably does not adhere with the HFEA Code of Practice's stated factors which are to be considered when assessing the appropriateness of PGD applications. Indeed, due consideration of the evidence given in the relevant literature about the capacities and quality of life possible for persons living with Down's syndrome would seriously call into question the validity of a positive judgment recommending PGD as a treatment service for Down's syndrome according to the current UK regulatory instruments. I end the thesis by considering why the HFEA's relatively recent decision to have an exclusive list of "serious" and therefore "in principle" test-worthy conditions stands to entrench prejudice, stigma, social bias, and unfair discrimination against the disadvantaged social group of persons living with Down's syndrome.

The advent of PGD has meant more choices—often experienced as very difficult ones [68]—for some parents in determining the outcomes of reproduction. More than ten years ago the Joint (HFEA and Human Genetics Commission) Working Party recommended "placing greater emphasis on the role of those seeking [PGD] treatment in reaching the decision about when treatment [is] appropriate, whilst at the same time maintaining that this should not imply that this treatment should be available on demand".[29:5, para.23] Admittedly, the standard of "seriousness" is unlikely to whither soon insofar as it is still viewed as a stop-gap against PGD being "available on demand". As we shall come to see, the HFEA has resisted any definition of "seriousness" and there has been quite a bit of concern expressed in both professional and popular writing to establish where the line should be drawn to be able to sort what is from what is not a serious condition. Elsewhere I have begun work to show that line-drawing according to "seriousness" is value-laden and context-dependent so that, as Elisabeth Gedge puts it, "a principled approach to the regulation of PGD" according to this standard is "difficult to envision".[69:248] My tack with *this* thesis is not to take on the general question of what makes a "serious" condition distinct from a non-serious condition. My original contribution to the field here is in beginning the work needed to assess the *ethical implications* and the *supportability* of this standard and the regulatory approach surrounding it by way of analysing the case of Down's syndrome.

Strategically, the thesis is set up to influence policy by working from within the standards for assessment given by the UK regulatory framework, first running the test case that then ends up pointing us back to reconsider why and what form of regulation of parental choices with respect to PGD are needed—a question we will not manage to avoid by implementing the "seriousness" approach. Admittedly, the test case, with its dependence upon empirical evidence, will be open to challenge, especially given the vague standards employed in the relevant legal and policy instruments. Be that as it may, the thesis will provide an example of the depth made available through a recent naturalized turn in bioethics,[70-72] thus insisting on grounding debates concerning the differential impact of disabilities in empirical evidence. As such, it puts into practice what some disability scholars have complained of as missing from philosophical (including bioethical) discussions of disability, and that is an appreciation of disability in context, not as one thing, but rather as a complex of interrelations—an outcome of an interaction between features of people and features of the physical, social, and attitudinal environments.[73-75] Disability studies, bioethics and philosophy of law scholar Jerome Bickenbach has written that: "Conceptual confusion and social policy incoherence result … when the complexity is ignored and the entire notion [of disability] is reduced to one component feature of the interaction, be it the biomedical or intrinsic aspect or the social or socially-constructed aspect".[73:49] Accordingly, I think that answering the question of whether Down's syndrome should be considered a "serious" condition (for which PGD would therefore be indicated as an appropriate treatment) needs to be answered in a way that respects the complexities involved across the diverse components (e.g., biological, psychological, social, economic, cultural, environmental, ecological, etc.) and their dynamic interrelationships given in the *lived experience* with disabilities for members of this population.

Structure

This thesis is structured in three parts. Before beginning the critical analysis of the specific case of regulating PGD for Down's syndrome, it is important to understand the formative context within which the specific regulatory instruments under investigation have the meaning in practice that they do. Chapter one provides a snapshot of the very influential Warnock Report in order to situate the historical context for policy discourse on regulating ARTs in the UK. This discussion provides important background for understanding the genesis of the relevant laws and Code of Practice which would eventually be used to regulate PGD in the UK. In this chapter I also set forth some of the main theoretical commitments of power-focussed feminist bioethics as my chosen approach to the materials: an approach that motivates filling in some of the moral blind spots that are part of the continuing legacy of the Warnock Report. Chapter two begins with an explanation of the specific legal and regulatory instruments as well as the ethical rationale for the HFEA's protectionist approach to PGD regulation. I then outline the factors stated in the Code of Practice that are to be considered by attending ART professionals when deciding the appropriateness of any PGD application. I next explain evidence from a variety of relevant scholarly literatures which would have a bearing for each of the factors used to index judgments concerning the appropriateness of PGD for Down's syndrome. Given that my assessment of the relevant evidence would challenge a judgment in favour of the appropriateness of using PGD to prevent birth of children with the disabilities associated with Down's syndrome, I pause in my argument to consider counter-arguments of this assessment by discussing two prevalent models for understanding disability: namely, the social versus medical model of disability and how they argue for different views on whether Down's syndrome is a "serious" condition and if it is, what makes it so. Chapter 3 begins by explaining why having Down's syndrome on the HFEA's list of exclusive, "in principle", "serious" conditions is misleading and therefore harmful to persons living with Down's syndrome and potentially their families as well. Chapter 3 then draws from some of the lessons learned from investigating the particular question of whether or not Down's syndrome should be a candidate on the HFEA's list of conditions for which PGD testing is exclusively permitted and explains some broader reasons to expunge the list altogether.

Chapter 1: Matters of context and approach

Before starting my analysis of PGD regulation in the UK, some preliminary remarks are in order to situate the theoretical mainsprings and historical context of this discussion. As such, the purpose of this chapter is, firstly, to provide some historical background through a brief analysis of the basic rationale behind the Warnock Report on new ARTs, highlighting the confining effect it has exerted on the ensuing public discourse of the ethics of ARTs. Next I explain some of the wider ethical considerations as raised by power-focussed feminist bioethicists for attending to, sometimes unmasking, and evaluating the oppressive effects of law, regulation, and social policy for disadvantaged social groups. In sum, this approach would seem especially useful for filling in some of the moral oversights concerning the regulation of new ARTs in the UK as left in the wake of the Warnock Report.

Historical background: the Warnock Report

In 1982 a UK parliamentary committee of inquiry was convened under the directorship of philosopher Mary Warnock, with a mandate "[t]o consider recent and potential developments in medicine and science related to human fertilisation and embryology; to consider what policies and safeguards should be applied, including consideration of the social, ethical and legal implications of these developments; and to make recommendations."[61:para.1.2] Some of the innovative reproductive technologies the Committee dealt with included IVF, artificial insemination, gamete and embryo donation, surrogacy arrangement for pregnancy, trans-species fertilisation, and the use, manipulation, and destruction of human embryos for the purposes of scientific research.[76] PGD was not yet developed as a clinical application at this time, but the Warnock Committee's eventual recommendations would importantly inform legislation and regulation that would eventually be used to control PGD licensing.

Until then, research with human embryos and the development of new reproductive technologies had evolved "in an unstructured and unmonitored way" and the Warnock Committee concluded that "[t]he interests of those directly concerned, as well as those of society in general, demand[ed] that certain legal and ethical safeguards should be applied."[61:para.13.2] The committee found that the consulted public(s) were generally agreed that some legal barriers "must be set up", but the British public (as a whole) was quite fractured and without agreement on "where these barriers should be placed."[61:sec.8] Warnock summarized the situation thus:

> Moral questions, such as those with which we have been concerned are, by definition, questions that involve not only a calculation of consequences, but also strong sentiments with regard to the nature of the proposed activities themselves.[61:Fwd, para.4] ...In recognising that there should be limits, people are bearing witness to the existence of a moral ideal of society. But in our pluralistic society it is not to be expected that any one set of principles can be enunciated to be completely accepted by everyone. This is not to say that the enunciating of principles is arbitrary, or that there is no shared morality whatever. The law itself, binding on everyone in society, whatever their beliefs, is the embodiment of a common moral position. It sets out a broad framework for what is morally acceptable within society. Another philosopher put it thus: "The reasons that lead a reflective man to prefer one ...legal system to another must be moral reasons: that is he must find his reasons in some order of priority of interests and activities, in the kind of life that he praises and admires". In recommending legislation, then, we [the Committee] are recommending a kind of society that we can, all of us, praise and admire, even if, in detail, we may individually wish that it were different.[61:Fwd, para.6]

Interestingly, philosopher and public policy critic, Will Kymlicka has observed that, in the main, the public disagreements over new ARTs have not been concerning how to weigh conflicting principles; instead, most have been of the " 'slippery slope' variety".[77:254-255] On the one hand, certain parties contend that new ARTs over time will degrade human reproduction and the status of women, upset the "family unit", lead to social intolerance of citizens living with disabilities, and serve eugenic purposes. On the other hand, defenders of new ARTs emphasize that such outcomes are not inevitable and that regulation can prevent most, if not all, potential abuses.[78;79] According to Kymlicka, these disagreements across the public are deep, but do not indicate disagreements over values. In this regard, his assessment might seem to be somewhat of an oversimplification—though surely, Kymlicka's phrasing is rhetorical rather than literal—when he explains this, saying that: "Everyone agrees that eugenics is wrong, that prejudice against women or the disabled is wrong, that family instability is undesirable, etc. They simply disagree about whether [new ARTs] will have any of these implications…"[77:254-255] Assuredly some arguments concerning new ARTs of the "slippery slope" variety are put forth strategically for pushing partisan concerns and do not really count as warnings or projections founded on genuine beliefs concerning impending developments or consequences. Still, public disagreement over new ARTs, according to Kymlicka, remains largely empirical and the prevalence of slippery slope arguments in this arena indicates, at one level, *appeals* to some sort of shared values[77:267, nt.24]—or, perhaps, a hope for the creation thereof.

As such, Kymlicka surmises that public disagreements over new ARTs commonly reflect differences in power as seen by the fact that some of those parties most serious about the implications of new ARTs—e.g. disability rights activists, women's groups, environmentalists, and public health advocates—are those that feel they have little or no say over the future research, development, service delivery, rules of access, and other facets involved in directing the course of these technologies.[77:254-255;see 80] If we link this insight back to the Warnock Committee's contention (given above) that how we seek to control these technologies reveals various moral ideals for what kind of society we wish to protect and promote (as expressed in and through law[see 81]), it makes sense that choosing how to regulate new ARTs has been the site of competing social, political, and ethical interests with various "voices" vying for public recognition and influence over the process. In this context, as Kymlicka aptly points out, "[t]aking morality seriously, in the first instance, requires taking people seriously—showing concern for people's lives and interest[s]."[xi;77:251]

Historically speaking, the Warnock Committee operated with a keen awareness of its pragmatic function, knowing that its recommendations were needed as a basis upon which to draft legislation over new ARTs that would have to be binding on all citizens whether or not all citizens happened to agree with the ethical rationale behind the laws.[82:505] In providing the groundwork for regulating new ARTs and recommending where barriers to its uses should be placed, the Warnock Committee owned that not only were they engaging the question of "what kind of society can we praise and admire", but also "[i]n what kind of society can we live with our conscience clear?"[61:sec.8] This could be read as a commitment to *taking morality seriously*, not just an attempt to design regulation that would be socially acceptable to a large majority of the public. If taking morality seriously means showing concern for people's lives and interests (*à la* Kymlicka[77:251]), this forces the two-pronged question: "Concern for which people's lives and for which of their interests?" And, if we follow Kymlicka's read of the situation further, taking morality seriously in the design of laws and public policies to regulate new ARTs means, at the very least: i. identifying which people are affected by new ARTs; and, ii. "ensuring that

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[xi] Kymlicka notes: "More accurately, morality requires taking sentient life seriously, human or animal."[77:265,nt.17] To put a finer point on it, Kymlicka might more accurately have said human and non-human animals.

[new ARTs] are used in such a way as to promote, or at any rate not to harm, their legitimate interests."[77:251]

It is difficult to speak (sensibly) of embryos themselves as having interests,[xii] and in UK law they have never been accorded protections in the form of legal rights. Still, the Warnock Committee asserted that "because of the special status that we [as a society] accord to the human embryo", research with embryos "must be subject to stringent controls and monitoring."[61:para.11.18] The Committee therefore strongly recommended that "research conducted on human *in vitro* embryos and the handling of such embryos should be permitted only under licence."[61:para.11.18] Given that PGD involves the handling of embryos (*in vitro*), and given that when first introduced as a clinical application in the 1990s PGD was considered (and remained so for more than a decade thereafter[84:233]) an experimental procedure, various parties in the UK who took a special interest to try to influence policies pertaining to human embryo research also saw their interests at stake in policies that would govern PGD and other fertility treatment services or *vice versa*.[xiii;cf. 78;87;88] The politics and ethics of human embryo research in the UK have been well documented, and have included most significantly proponents of scientific research—with protectionist moves on the part of the medical establishment—often in contestation with spokespersons of the religious right.[89] These veritably warring (interested) parties have dominated the relevant public debates not only with respect to the ethics of regulating human embryo research but also the ethics of fertility services and new ARTs.[80]

Pressure on the Warnock Committee [cf. 82:505 & 521] to come up with a national consensus (under government auspices) on which to base legislation meant that the interests of certain social groups who lacked political influence were overlooked: obviously, some interested parties lacked the means to make their worries and/or dissenting voices heard.[80:99] Accordingly, the Report came out in 1984 and failed to consider the impact of new ARTs on children, persons living with disabilities, and women[77:254]—not surprisingly, all politically and

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[xii] Some would claim that embryos are potential interest bearers. Though I will not argue whether or not thinking of embryos as potential interest bearers makes sense, I should say that it would be difficult to defend such interests as having any sort of parity with the interests of women who are interested to become mothers, or couples interested to become parents. Presumably this view of embryos accords with the fact that they are not accorded rights under UK law. See discussion in de Wert and Mummery.[83]

[xiii] Sourcing human embryos for research has always proven challenging and, not surprisingly, to this end scientists have turned to fertility clinics for supplies of supernumerary embryos.[85] Since PGD is one kind of test used to determine which embryos are implanted and which ones will be left over in the context of *in vitro* fertilization, there are various ways in which rules governing access to PGD and what applications are made licensable could affect the supply of supernumerary human embryos and therefore count as potentially impacting on human embryo research.[86]

socially marginalized social groups[90] with comparatively fewer resources to make their voices heard.[80] The Report identified "the value of human life"[91:xvi] as its most significant issue. Later, in a published reflection on the Committee's mandate, Warnock stated that she considered all other issues comparatively "trivial"[xiv] when measured against the importance of questions related to human embryo experimentation[77:266,nt.23;91:xvi;92]—"a view", Kymlicka charges, "that surely reveals a certain moral blindness."[77:254]

In framing the matter of new ARTs in this way, the Report has been read by some feminists as having had a polarizing effect on the subsequent public discourse of the attendant ethical, social, and legal issues. As feminist Edward Yoxen has observed, the ensuing debate increasingly focused on " 'an over-individualized notion of the embryo' that ignore[d] the site of its origins and the necessary conditions for its subsequent nurture and development."[80:95;93] In effect, the carrying women were reduced to near invisibility.[cf. 94] At the time, political scientist and founder of the International Reproductive Rights Research Action Group, Rosalind Petchesky, charged the Report with underwriting a "lopsided preference" for "embryonic life over the lives of women patients", thus demonstrating "a wilful disregard of patient interests".[95:xv-xvi] Feminist philosopher Anne Donchin has pointed out that the debate over new ARTs was now cast in terms of the moral status of the embryo as an autonomous entity.

Motivations to redress this moral context as per feminist approaches to bioethics

With PGD being one of the "newer" of the new ARTs, much of the on point moral-cum-political discussions in the scholarly literature have understandably taken the form of either ethical surveys which attempt to remain politically neutral, or more partisan commentaries often defending an exclusive set of interests or values. The interests most commonly defended in this literature, as well as in UK law and policy, have been those related to protecting the welfare of the child-to-be[xv]—what has been described as the HFEA's "moral bedrock"[97:421]—with consequences for how we are obligated to treat embryos. It is my view that many of the

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[xiv] Kymlicka notes that Warnock, in reflection on the Report, noted that moral questions having to do with new ARTs are of two kinds: "'the first kind centers on the concept of the family; the second on the justification, or lack of it, for research using human embryos. My [i.e. Warnock's] personal belief was, and is, that the second set of questions is both more important and more difficult than the first.'"[82:506] Kymlicka further quotes Warnock as having said that: " 'All the other issues we [the Committee] had to consider seemed relatively trivial compared with this one' "[91:xvi] (as quoted by Kymlicka).[77:266,nt.23]

[xv] Lately there has been a move to give greater acknowledgement to the interests of families.[96] We will discuss this further in the next chapter.

relevant interests of various persons living with disabilities—as with the Warnock Report—have been under-represented in public as well as scholarly research discussions. To redress this lacunae in the literature, I contend that a feminist bioethics [70;98:4-5] lens [99] is especially useful for making visible whose interests are being overlooked or adversely affected, and whose interests are being protected or promoted, through the legal and policy instruments regulating PGD in the UK.

Understanding the "actual particulars of the encompassing dominance structures"[100:52] and the dynamics that make possible or entrench oppression have been historically key to feminist ethical analysis, and by extension to contemporary feminist bioethical analysis.[101-104] Feminist bioethics is an umbrella term. One influential stream, power-focussed feminist approaches to bioethics, takes as its primary goal to abolish (or, at least significantly modify) any system, structure, or set of norms that contributes to the oppression of disadvantaged social groups.[103:38] In this respect, feminist philosopher Margaret Urban Walker has argued (with others[90;100;105]) that "[t]he most obvious way feminist ethics and politics connect morality and power is in examining the morality of specific distributions and exercise of power."[71:4] This is to say, a power-focussed feminist ethics looks at "how power is distributed equally and unequally, whether and how it gains legitimacy or sustains itself illegitimately, and whether and when people [or organizations] having power over others is, in Walker's words, 'morally necessary, arbitrary, or catastrophic.' "[xvi;105:x] A power-focussed feminist approach to bioethics, then, demands that we take seriously the role that health law and public policy assume in relation to general structures of oppression[xvii] in society. This means that in the context of PGD, regulation affecting clinical access needs to be critically examined not only in terms of the effects on those individuals directly involved—namely, AHR clients and AHR professionals; it must also be

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[xvi] Moreover, feminist anti-oppression theorists are especially concerned to understand inequalities deriving from problematic relations and dynamics between social groups, not simply between individual members within given groups.[90;106:83] In making the investigation of group-based power differentials central to the agenda of doing feminist bioethics, feminist philosopher and bioethicist Susan Sherwin proposes: "that we always interrogate [health] practices or policies to determine how each contributes to existing patterns of oppression, especially as they affect women: does it worsen or exacerbate current inequalities? Is it likely to improve the situation? Or is it neutral with respect to social group differences? As well, we need to ask whether there are alternative practices or policies we might adopt that would make a larger contribution to reducing social inequalities."[106:84]

[xvii] Marilyn Frye defines oppression as consisting in "an enclosing structure of forces and barriers which tends to the immobilization and reduction of a group or category of people."[107:10-11] Sally Haslanger and colleagues explain that: "Not just any 'enclosing structure' is oppressive, however, for plausibly any process of socialization will create a structure that both limits and enables all individuals who live within it. In the case of oppression, however, the 'enclosing structures' in question are part of a broader system that asymmetrically and unjustly disadvantages one group and benefits another."[101:n.p., online]

assessed with respect to the surrounding interlocking patterns of bias, prejudice, and unfair discrimination against relevantly affected social groups.[98;106] In this regard, there is overlap between power-focussed feminist approaches to bioethics and critical disability studies insofar as the former seeks to identify and expose[cf. 108:217-218] not only sexist, but also ableist implications of laws, regulations, guidelines, and public policies which might otherwise appear as neutral or objective.[xviii][103:90]

Certain consequentialists (and defenders of the status quo in the UK) might rest easy with the fact that PGD, after all, directly affects only a very few, and often much advantaged (e.g. educated, wealthy) set of clients. Highlighting that the HFEA is, after all, only an embryology authority, they might plead that responsibility for the wider effects of prejudice, stigma, and discrimination against persons with disabilities is a much bigger issue in which the HFEA's part is very small. In contrast, by drawing upon the anti-oppression goals of feminist bioethics this thesis is provided with an impetus to look for how enclosing structures of forces and barriers underwritten by the regulatory regime for PGD in the UK form a part of a broader system that asymmetrically and unjustly disadvantages the group of persons living with Down's syndrome and their families as well as other relevantly related groups of persons living with other disabilities. As Sherwin underscores: "If we want moral change and not mere moral platitudes, then the particular practices that constitute oppression of one group by another must be identified and subjected to explicit moral condemnation; feminists demand the elimination of *each* oppressive practice [emphasis added]."[98:54]

Much work in feminist bioethics is also critical of the limitations imposed when restricting (or just leaving) ethical analysis at the level of abstract and general principles[100;110:2]—a predominant approach in mainstream bioethics.[104;see 111] Instead, many feminist bioethicists have stressed the paramount importance of context[cf. 112]—including the situated, relational spaces in which ethics is practiced—to determine and adequately respond to actual, real-life situations "that morally concerned persons find problematic".[98:76] From a power-focussed, feminist bioethics perspective, as Sherwin explains, "the relevant concrete details to be considered in our ethical deliberations include the political or power relations of the persons who are involved in or who are affected by the practice or policy being evaluated."[100:51-52]

As such, an important strain within power-focussed feminist bioethics, also influential as one of the mainsprings directing the approach to the materials in this thesis, is feminist relational theory. Feminist relational theory is again an umbrella term designating a range of

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[xviii] See Bartlett (1993)[109:551] and Sherwin (1992).[98:4-5]

perspectives. According to Catriona MacKenzie and Natalie Stoljar's very influential introduction, "[t]hese perspectives are premised on a shared conviction, the conviction that persons are socially embedded and that agents' identities are formed *within*" [113:4] and *through*[114:4] the context of social relationships, shaped, as it were, by a complex of intersecting "socially salient features of their embodied lives", such as race, class, gender, ethnicity, age, and disability status.[115:10] As feminist bioethicists and legal academics Downie and Llewelyn explain: "The central question in each case is not so much 'what is *X* in relationship to or with?'—an approach, I would add, ascribed by relational theorists to liberal individualist atomistic theories of identity[116]—"but, rather, 'what is the effect of being in relation?'"[114:4] Feminist relational theorists who hold to the social embeddedness of selves are apt to question whose interests are served and whose are harmed by conventional structures of thought and practice: they are concerned to emphasize the important effects that social pressures (often implicit and structural in nature) have on persons', and even groups', autonomy and moral and political agency.[xix;113:4;117-119]

There are, of course other theories which see the constitution of personal identity *within and through* one's social relationships, but feminist relational theory importantly focuses on how some of the social determinants that contribute integrally to our identities can be pernicious, as with the effects of oppression, which sometimes thwart or damage one's self-esteem, self-trust, and autonomy. By pointing out the differential impact that such forces can have on personal development, functional capacities, and access to opportunities, feminist relational theorists show not only that our identities are socially constructed and conditioned: they also show, as Canadian feminist bioethicists Baylis, Kenny, and Sherwin explain, that "'we are not all constructed as equals'".[120:201] As such, feminist relational theorists tend to invoke social justice concerns in looking at the context in which political and social structures are generated and sustained, and through which law and policy decisions are designed and implemented. As Baylis, Kenny, and Sherwin further add,

> [social justice] asks us to look beyond effects on individuals and to see how members of different social groups may be collectively affected by private and public practices that create inequalities in access and opportunity. In this way, social justice reflects our relational understanding of persons as socially constituted and situated. Social justice

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[xix] In applying this theory to ethics, the focus of relational approaches across several feminist bioethicists followed in this thesis, is to "analyze the implications of intersubjective and social dimensions of selfhood and identity for conceptions of individual autonomy and moral and political agency."[113:4]

> further enjoins us to correct *patterns* of systemic injustice among different groups, seeking to correct rather than worsen systematic disadvantage in society.[120:203]

The impetus voiced in the introduction to this thesis to service a more holistic, non-reductionist appreciation of the disabilities[xx] associated with the lived experiences of persons with Down's syndrome and their families is what propels my choice for approaching the materials through the lens of feminist relational theory. The emphasis of this theory on making sense of individual well-being, not just according to factors intrinsic to the individual but also extrinsic factors arising from the wider, socially embedded context in which she finds herself, seem to track with many of the specific factors for assessing the appropriateness of PGD applications given at section 10.7 of the Code of Practice. For example, appreciation for the relational dimensions, situational context, social, and (as we shall see) even historical background are key for assessments that would gauge "the likely degree of suffering associated with the condition", "the social support available", and "the family circumstances of the people seeking treatment"[121:s.10.7]—all factors critical for appreciating the complexity of the involved contingencies of associated disability for a life with Down's syndrome.

My choice of feminist relational theory as a lens for analysing the ethics *in* regulating PGD has several further motivations: firstly, it allows for a social critique when much of the bioethics literature on the topic takes the individual client or patient as its primary starting point. Secondly, relational theory requires looking at, not only how individuals relate to and affect one another, but also how group status and dynamics affect moral agency and moral outcomes. There is a great deal of research that is still needed to make sense of the complexity of the ethics *in* regulating PGD; still, some does already exist which documents the voices, and a lot exists that analyzes the *experiences*, of relevant disadvantaged minority groups, including the various disability populations and their families. The voices of the members of these groups as well as studies of their own and their families' experience are still at the margins of philosophical, bioethical, and legal discussions of how to regulate PGD.[xxi] In drawing upon feminist relational theory as a lens to focus the ethics embedded (and at stake) in the regulation of PGD in the UK, it is my hope that doing so will motivate an interdisciplinary and naturalized approach to the material which stands a good chance of not just being original but also more

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[xx] For an influential spokesperson for the Nordic relational theory of disability see Jan Tøssebro as summarized by Shakespeare.[75:25-26]

[xxi] Indeed, with respect to the realities of living with Down's syndrome and parenting a child with Down's syndrome, much of the on point social science literature seems under-utilized in the relevant medical and social policies pertaining to prenatal testing and care.

accurate, comprehensive, and in effect responsible to the lived realities faced by persons with Down's syndrome, their families, and other relevantly related social groups of persons living with disabilities.

Contemporary feminist philosopher and bioethicist Samantha Brennan contends that the feminist approach to doing ethics commonly seeks inclusion and respect for different moral perspectives "not through universalization but rather through detailed attention to particulars."[105;108;112;122:861] Although it is commonly agreed for these scholars that in theorizing a methodology for bioethics, morality cannot be distilled into a set of abstract, universal rules "which can be specified apart from their context of use," [98:77] this does not mean that principles are necessarily unimportant. Indeed, power-focussed feminist bioethics often encompasses theories that stake acute moral (and political) concerns based on the assertion of principles of social justice[115;120;123;cf. 124]—this, as mentioned above, follows from the demands for criticism of patterns of oppression within, and across, different social groups. But justice—and social justice too, according to most power-focussed feminist bioethicists—cannot be made sense of in the abstract. As Sherwin explains in *No Longer Patient*,

> [t]o speak meaningfully about justice, it is necessary to examine the actual forces that undermine it, as well as those that support it[98:55]. ... [J]ustice is defined in terms of its opposition to oppression. The principle [when appealed to] only makes sense when the relevant contextual details are spelled out. Oppression is not a phenomenon that can be adequately explored in the abstract; contextual details about the specific form of oppression and about other relevant features of the situation in question must be added to make sense of the moral concerns raised.[98:82]

Though there are debates within feminism as to whether a focus on gender and women's experience is a necessary condition to qualify any program as feminist, [122;125:146;cf. 126:95] the relational school in bioethics with its anti-essentialist platform insists upon the real diversity of women's experience across its intersectional dimensions and that fighting oppression means fighting sexism, ageism, racism, classism, and ableism for any group of oppressed social groups, even if most particularly for women.[102;cf. 127] My project does not argue for, but rather takes as a starting point feminist bioethicist Susan Sherwin's assertion that: "The principal insight of feminist ethics is that oppression, however it is practiced, is morally wrong. Therefore, moral considerations demand that we uncover and examine the moral injustice of actual oppression in its many guises."[98:54]

Clearing a space from within

There is one further reason I want to put forward for my choice of a "ground-up" approach[cf. 128] to the materials used to build this thesis. How we choose to have our children, and how we choose what kinds of children might be worth preventing from being born, are questions over which there has historically been a lot of social agreement in UK society. More recently, UK society is much more fractured in its moral standpoint(s) on these issues since the inception of the new ARTs, which include PGD. I do not presume that the fact of consensus is necessarily a sign of positive moral worth, and there is much in the moral terrain of critical disability and gender studies that shows that social consensus can be morally pernicious. But that being said, legislation in a democratic society, as Warnock pointed out, has as a reasonable goal not to produce unanimous agreement on its substance from all citizens but (minimally) the law needs to delineate that to which "everyone would be prepared to consent".[82:521] Experience from the history of bioethics shows that building moral consensus across people of diverse backgrounds with different ethical orientations is more likely when discussions over how to do so proceeds in a case-by-case manner, showing respect for particularity, concreteness, reflection on experience, and dialogue rather than trying to ground specific judgments in general, foundational principles.[98:79;129] As such, this thesis does not start by trying to dismantle the standard of "seriousness" nor the list of (exclusive) conditions deemed serious by the HFEA. Instead, it circumscribes its starting point to a discussion of the suitability (or appropriateness) of using PGD as a treatment service against Down's syndrome through a concrete investigation of the evidence that can be brought to bear on this case as called for according to current UK law and policy.

As John Locke says in his Epistle to the Reader of his *Essay Concerning Human Understanding*, "it is ambition enough to be employed as an under-labourer in clearing the ground a little, and removing some of the rubbish that lies in the way to knowledge."[130] Following in the spirit of Locke, much of the critical discussion to follow in this thesis is an attempt to point up and begin to clear out what might otherwise stand in the way of sound regulatory law and policy—law and policy that could, perhaps, be more ethical than the present UK regulatory regime and more *deserving* of public support. Using this as an overall goal for the course of this thesis and following some of the main lines of approach defended by feminist bioethics, I take as my starting point a thickly descriptive discussion of the present law and policies for regulating PGD in the UK, only then moving onto a normative assessment of the regulatory instruments and system as a whole.[cf. 131]

Chapter 2: Is PGD for Down's syndrome appropriate according to UK law and policy?

Chapter objectives and outline

As noted in the last chapter, the Warnock Report warned of ethical risks at stake with the new ARTs and consequently recommended legal restrictions of their use. The legislation to follow attempted to speak to the worries of a range of interested parties and to protect against various projected harms with varying degrees of success.[132;133] It is the overall burden of this thesis to show some of the main problems with the UK instruments for regulating PGD in terms of their built-in negative potential for harms against persons living with Down's syndrome. The purpose of this chapter is twofold: 1. to explain the instruments used to assess whether or not an application for PGD is considered appropriate according to UK law and policy; and, 2. to determine whether or not the application of PGD for trisomy 21 (the chromosome implicated in Down's syndrome) is an appropriate application of this technology as per the UK instruments for assessment. Referencing the HFEA's own standards for licensing and assessment of the legitimacy of PGD applications, I will argue that the justification for extending PGD testing for Down’s syndrome (as a permissible use of this technology) under the current UK legal and regulatory instruments does not follow from a consideration of the evidence as given in the relevant scholarly literature.

As to the stages of my argument, I begin with a short explanation of the Human Fertilisation & Embryology Act [HFE Act (1990)] and the Human Fertilisation & Embryology Authority (HFEA) as, respectively, the statutory foundation and executive body of authority which are the practical basis for PGD regulation in the UK. I then outline the factors to be considered when deciding the appropriateness of any PGD application as given in UK law and policy. Next, I argue that licensing PGD testing for Down's syndrome (not in itself, but) on the basis of these factors is problematic. I end by addressing some counter-arguments of this assessment through a general discussion of the medical versus social model of disability for making sense of what does or does not make Down's syndrome a "serious" condition.

The HFEA Act (1990) and the HFEA

In 1990 the Human Fertilisation & Embryology Act was passed as an Act of Parliament of the UK; as stated before, the Act was designed on the basis of the Warnock Committee's recommendations. The prepared report of the Warnock Committee judged that:

> The protection of the public, which we see as the primary objective of regulation, demands the existence of an authority independent of Government, health authorities, or research institutions. The authority should be specifically charged with the responsibility to regulate and monitor practice in relation to those sensitive areas which raise fundamental ethical questions. We therefore recommend the establishment of a new statutory licensing authority to regulate both research and those infertility services which we have recommended should be subject to control.[61:sec.13.3]

The Act (1990), therefore, created the HFEA, an executive, non-departmental public body—the first statutory body of its type in the world—that officially started work on 1 August, 1991.

Over the past two decades, the HFEA has secured for itself a robust national and international reputation owing in part to its "accessibility to the public and its openness in communicating information about its activities" which has "contributed to the general acceptability of the [UK] regulatory framework and international interest in adapting its outlines to conditions in other countries."[80:95-96] The HFEA is also somewhat unique for the fact that it not only has a statutory foundation for AHR service delivery and research; it also has a developed set of regulations and guidelines in the HFEA Code of Practice. Moreover, in the arena of embryo ethics and politics—an arena that is commonly very divisive across various stakeholders[78;89]—the UK approach is understood as a moderate, workable regulatory framework situated between the extremes of highly restrictive regimes such as Germany [134] and Italy [135] which maintain a statutory ban on PGD, and highly permissive regimes such as the USA[97] which has neither legislation nor national policies to regulate PGD.[136;137] For these reasons, the HFEA is acclaimed as a kind of *beau ideal* of ART regulators and as such enjoys considerable international influence.

How to determine the permissibility of any PGD application

Clinical applications for PGD were originally developed at the end of the 1980s[5;138;139] as an alternative to prenatal diagnosis[140;141] (PND) followed by elective termination for fertile clients at reproductive risk of passing on a heritable genetic condition to their prospective offspring.[142]

In a very influential article entitled, “Genetic screening and ethics: European perspectives” a consortium of authors lead by Ruth Chadwick have written that: “There is a consensus that the purpose of prenatal testing is to detect serious diseases and that, despite varying degrees of stress on individual autonomy, there does not exist a right to test for everything.”[50]

The original HFE Act (1990) did not either directly address PGD or prescribe specific conditions for its use. Nonetheless, the Act (1990) accorded the HFEA discretionary authority to issue licenses for activities involving the handling and/or manipulation of embryos: i) to provide treatment services; ii) to store gametes and embryos; and iii) to carry out research. Under UK law, all of these activities involving human embryos are prohibited unless performed with licensed authorization.[143:169] As such, PGD has been, and still is, licensed as a "treatment" service.[144:Sch. 2, s1(3)] And, as a treatment service, the HFEA continues to authorize PGD as a practice "designed to secure that embryos are in a suitable condition to be placed in a woman".[xxii;144:Sch. 2, s1(1)(d)]

In a 1998 study which surveyed the results of the legal status and attitudes towards PGD from thirteen different countries, Viville and Pergament observed that: "Most of the countries with centres offering PGD, [including the UK], consider it an early form of prenatal diagnosis."[146:1378] After a public consultation in 1999, the HFEA and Human Genetics Commission Joint Working Party on PGD published (in 2001) an official set of principles for restricting the use of PGD, including the recommendation that affirmed that "guidance should state that indications for the use of PGD should be consistent with current practice in the use of PND."[xxiii;29:pt.26, rec.10] Historically speaking, this remained a guiding principle for embryo selection in the UK as per the Code of Practice (6th edition, 2003) which declared that: "indications for the use of PGD are expected to be consistent with current practice in the use of (post-implantation) prenatal diagnosis (PND)."[xxiv;149:s.14.21] This confirmed that which the HFEA

[xxii] As Johnson and Petersen note: "the extent of parental genetic choice is restricted to the interpretation the HFEA places on 'suitable.' "[145:725]

[xxiii] Again, according to a 2000 HFEA *Consultation document on PGD*: "Both PND and PGD raise the same general issues in relation to the seriousness of inherited conditions."[6:pt.27] See also HGC (UK) and Albert[22:pt.9] and HFEA.[147:11]

[xxiv] This principle was dropped with the Code of Practice (7th edition) in 2007. The issues involved here are very complex and touch on distinctions between regulation concerning women's rights to information relevant for elective termination of an affected pregnancy versus women's rights to information relevant for initiating a pregnancy with unaffected embryos. PND involves testing *in vivo* whereas PGD involves testing *ex utero*/*in vitro*, and arguably does not invoke a woman's rights to sovereignty over her own body—or, at least, not to the same degree—as with cases of elective termination. The latter principle could be invoked to argue for more permissive access to PND as compared to PGD. On the other hand, in the European context especially, there has been patient protest against more restrictive access to PGD as compared with access to PND on the grounds that harms to

Advisory Committee on Genetic Testing had already established in declaring that "where the suitability of PGD is being considered, centres are understood to be applying the criteria for termination of pregnancy for foetal abnormality published by the Royal College of Obstetricians and Gynaecologists (RCOG). This limits the use of PND followed by elective termination to cases where there is a precise diagnosis and a 'substantial risk' of a 'serious handicap.'"[xxv;6:pt.34] Interestingly, a definition of "serious" is not given by the RCOG.[xxvi]

Under the current HFEA Act (1990 as amended in 2008[xxvii]), several new clauses now "place onto the face of the legislation the purpose for which embryo testing can lawfully be carried out."[155:5] Accordingly, "in a case where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality", the HFEA is authorized to grant a license to the relevant applicant (*i.e.* clinic) for the purpose of "establishing whether it [the embryo] has that abnormality or any other gene, chromosome or mitochondrion abnormality."[144:Sch. 2,s1ZA(1)(b)] It is now a statutory requirement[156:s1.1-1.4] under the Act (as amended 2008) that a license for the aforementioned purpose cannot authorise the testing of embryos,

> unless the Authority [HFEA] is satisfied – (a) in relation to the abnormality of which there is a particular risk, and (b) in relation to any other abnormality for which testing is to be authorised under sub-paragraph (1)(b), that there is a significant risk that a person with the abnormality will have or develop a

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embryos are of less significance than harms to foetuses. These arguments can be seen to divide over whether one approaches regulation from the perspective of defending against risk of greater harms to the conceptus versus risk of greater harms to women's reproductive autonomy. So far as I know, this issue is very under-discussed in the literature. There is some discussion in Rao.[148]

[xxv] Currently, the Abortion Act 1967 (c.87) (1.(1)(d)) states that abortions on grounds of foetal abnormality are permitted provided "there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped."[150] To date there has been measured social support in the UK for PND and elective termination for what are thought to be more "serious" conditions. Even so, as Bonnie Steinbock notes, there is also a wide-ranging conviction that PND for the purposes of elective abortion for "trivial" conditions is morally unjustified.[36:179;151:105;152;153]

[xxvi] The RCOG has stated that a strict definition of what constitutes a "serious abnormality" is "impractical." In the College's own words, the reason that a "strict definition is impractical [is] 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."[154:n.p., online]

[xxvii] Human Fertilisation & Embryology Act 2008 (c.22). All references hereafter to the 1990 Act (as Amended 2008) will be to the following: Human Fertilisation and Embryology Act 1990. As Amended (2008) - an Illustrative Text (26 November 2008) <http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsLegislation/DH_080205> accessed 29 April 2013. The two pieces of legislation were meant to be read alongside each other. The illustrative text incorporates the two documents but has no official status.

> serious physical or mental disability, a serious illness or any other serious medical condition.[144:Sch. 2, s1ZA(2)(a-b)]

These mandatory requirements provide the grounding for the HFEA guidance in the Code of Practice (8th edition) which maintains that the "use of PGD should be considered only where there is a significant risk of a serious genetic condition being present in the embryo."[121:s.10.6] In the HFE Act (as amended 2008) most key terms are defined in the Preamble; however, the definition of "serious" is still nowhere given in either the Act or the Code of Practice (8th edition). As such, there is no standard for "seriousness" given by the HFEA.

What is "serious" about a "serious genetic condition"?

The HFEA is not at all unique in its failure to define (or directly address) the criterion of "seriousness." According to Wertz and Knoppers' wide reaching international survey, though "seriousness" is used as a cornerstone of regulating ARTs, the term "'serious' is never defined, nor do the laws or regulations specify who should be responsible for making a definition."[157:30] Wertz and Knoppers surmise that in doing so, law- and policy-makers are presumably relying upon a "common public understanding of what constitutes a serious disorder."[157:30] As such, this approach to "gate-keeping" access to ARTs, begets questions concerning the freedom of interpretation that consequently falls to attending ART professionals and their clients; it also raises issues concerning the legitimacy of legislative and/or ethics oversight.[xxviii;157:30]

In theoretical discussions (within and outside the UK) relevant ethicists, legal analysts, policy-makers, and clinicians have tended to distinguish between "serious" and "not-serious" (or "not-so-serious") genetic disorders on the basis of expected potential effects to quality of life.[48:133;148:67;163:608] But, the proposed quality of life measures for indexing seriousness have not

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[xxviii] Wertz and Knoppers have noted that: "Debates about what constitutes a serious condition are not unique to genetics. In the context of general medicine, seriousness is used to determine levels of services and government or private reimbursement. Yet 'serious' has proved impossible to define. The Institute of Medicine's Committee on Serious and Complex Medical Conditions [1999][158] examined possible quantifiable criteria, including international disease codes (ICD-9, DSM-IV, ICIDH-2), functional performance status scales [Karnofsky and Burchenal, 1949],[159] quality of well-being or activities of daily living scales [Katz et al., 1963; Lawton and Brody, 1969],[160;161] diagnosis-related groups (DRGs), diagnostic cost groups (DCGs) [Lamers, 1999],[162] utilization of services, physiological measurements, case management guidelines, and various combinations of the above. All were found lacking in one or more of the following: sensitivity, specificity, validity, or generalization to more than one disease. The committee concluded that 'the state of current clinical and research literature does not yet support the selection of specific categories of patients or service needs to define serious or complex medical conditions' (p.32)."[157:34-35] Wertz and Knoppers conclude, saying: "Policy makers cannot realistically expect to develop a universally respected definition of serious genetic disorders. The term—if used—must be qualified to include the judgment of the patient and physician, with the patient making the final, and one hopes, informed, decision."[157:35]

been successful at making firm designations concerning genetic disorders when moving from general considerations for assessment to the particulars of individual cases.[157:30;164] Others have tried to draw tighter lines to distinguish (what are described as) "serious defects" from "minor conditions" in the context of calls for the need to compile lists that would distinguish those genetic disorders that are "serious enough" to warrant PND. Dr. Jeffrey Botkin, defending an avowedly medical perspective, has proposed four criteria of assessment for legitimately adding a genetic condition to such a list: i. level of penetrance (i.e. the magnitude or risk of developing the condition); ii. age of onset; iii. effectiveness of treatment; and, iv. health impacts on the child and family.[60;157:30;165;166] In 1995, Botkin argued that Huntington's disease (late onset), foetal PKU (treatable after birth), and Tourette's syndrome (limited impact on life) are examples of "not-serious" genetic conditions and said that these would not warrant PND testing. In contradistinction, he put forth Fragile X syndrome, Cystic Fibrosis, and Down's syndrome as "serious" or "serious enough" genetic conditions and claimed that these conditions do warrant PND testing.[165] Still others have contested the frequency, if not the very practice (altogether) of drawing lines to define "seriousness." From a critical disabilities perspective, Adrienne Asch, Erik Parens, and Marsha Saxton have called attention to the designation of "serious" as socially constructed, reflecting the reigning biases and other forms of misperception germane to those dominant voices of the "temporarily abled" in our culture. According to these disabilities scholars and bioethicists, with the provision of adequate health services and social supports, what counts as a "seriously debilitating" condition—a judgment usually rooted in ableist misperceptions—need not be so, and almost always can be quite otherwise.[157:30;167-174]

Not surprisingly, the actual categorization of "serious" genetic conditions has proven highly indeterminate across relevant legal regimes, regulatory policies, and professional codes. Studies also show that in terms of clinical practice, what is judged to be a "serious indication" is highly variable across relevant geneticists and ART professionals.[157] As Knoppers, Bordet, and Isasi point out, the lack of commitment to a definition of "seriousness", coupled with an absence of consensus concerning what are examples of "serious conditions", means that "permitted uses of PGD based on a 'seriousness' test are likely to be interpreted very differently, which may create legal uncertainty for participants in the process, and may introduce both flexibility and arbitrariness in decision-making."[44:207]

However, the lack of a definite standard—this is to say, the lack of bright line to neatly divide instances of "serious" from "non-serious" conditions—though replete with certain of the liabilities named above, is not necessarily equivalent to having no standard at all. As a rule, conditions which are fatal, early onset, have a fast rate of progression, involve a great degree of suffering, and for which no therapeutics (or means of management) are available are generally seen to be more "serious" and less ethically contentious as applications for PGD (and PND) than conditions which are later onset, have a slow rate of progression, involve comparatively lesser suffering, and for which therapeutics (or means of management) are available.[xxix;36;175] Even so, at least since the time of the first UK public consultation on PGD at the turn of the millennium, there has been a call to include the judgment of the patient when assessing what counts towards a determination of "seriousness".[29] Hence, the Code of Practice (8th edition) states that:

> When deciding if it is appropriate to provide PGD in particular cases, the seriousness of the condition in that case should be discussed between the people seeking treatment and the clinical team. The perception of the level of risk for those seeking treatment will also be an important factor for the centre to consider.[121:s10.6]

The Code continues, also stating that in any particular case, the appropriateness of using PGD should be determined through consideration of the following factors:

(a) the views of the people seeking treatment of the condition to be avoided, including their previous reproductive experience
(b) the likely degree of suffering associated with the condition
(c) the availability of effective therapy, now and in the future
(d) the speed of degeneration in progressive disorders
(e) the extent of any intellectual impairment
(f) the social support available, and
(g) the family circumstances of the people seeking treatment.[121:s.10.7]

How to determine if PGD for Down's syndrome is appropriate according to the HFEA's standards for assessment

In using these factors as criteria for assessment, licensed clinical centres in the UK are given the responsibility to consider in which cases they can and in which cases they cannot justifiably offer[176:210-211] PGD services to clients. The HFEA has already approved PGD for Down's

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[xxix] PGD in the UK for quite some time now has not only been applied against examples of the former (e.g. Tay-Sachs Disease, Duchenne muscular dystrophy, Lesch-Nyhan syndrome), but is progressively on offer for what could be judged as "less serious" indications *according to these criteria* (e.g. certain types of Breast cancer (BRCA 1/2 genes), Colon cancer (HNPCC genes), and Early-onset Alzheimer's disease).

syndrome since the late 1990s.[177;178] Screening for foetal Down's syndrome first became possible in the late 1960s[179] and contemporary genetic testing procedures have been on offer as part of prenatal care in many modern industrialized countries, including the UK, since the 1980s.[180;181] Even if Down's syndrome is a condition that may not be considered (according to the previously mentioned criteria) as located at the extreme end of the spectrum of "serious" genetic conditions, it is nevertheless widely accepted (by professionals and the general public in the UK, and around the world for that matter) as a "serious" or "serious-enough" condition to warrant prenatal technological interventions (evidenced by longstanding, widespread public screening programmes[182-184]). It therefore is judged to be an ethically appropriate use of PGD (or PND).

Given that it is possible, and in the opinion of some desirable[65;67] to utilize PGD to select against Down's syndrome embryos in the context of IVF, is it appropriate for health care professionals to offer, and society to permit, the use of this technology for this purpose? What makes this condition so "serious"—in contradistinction to other "not-serious-enough" conditions—that PGD testing for it is deemed an acceptable use for this technology? Taking into account the HFEA's own criteria for making this sort of determination as given above with factors (a)-(g) but excluding (d),[xxx] I want now to investigate how it is that Down's syndrome qualifies as a "serious" condition for which PGD testing is to be judged appropriate. In what follows, I assess the relevant evidence that can be brought to bear for each of the above factors to determine the validity of such a judgment. I discuss in order:

1. the relevant reproductive experience and views of those seeking PGD for Down's syndrome [factor (a)];
2. the availability of effective therapies for Down's syndrome, now and in the future [factor (c)];
3. the extent of expected intellectual impairments associated with Down's syndrome [factor (e)];
4. the likely degree of suffering associated with the condition from the perspective of the individual with Down's syndrome [factor (b)];
5. the likely degree of suffering associated with the condition from the perspective of families to individuals with Down's syndrome, as well as how family circumstances and available social supports impact the expected suffering of all family members, including individuals with Down's syndrome [factors b, g, and f].

[xxx] Down’s syndrome is not a progressive disorder, and therefore factor (d) is irrelevant.

Assessing the appropriateness of a PGD license application

The views and reproductive experiences of PGD clients

> "(a1) the views of the people seeking treatment of the condition to be avoided,"
>
> "(a2) including their previous reproductive experience"[121:s.10.7]

In what ways could factor (a2), a couple's reproductive experiences, be seen as relevant for determining whether PGD for Down's syndrome is an appropriate application of this technology? Trisomy of chromosome 21, which leads to Down's syndrome, is one of the most frequent aneuploidies. But most cases of Down's syndrome are not inherited, and therefore being a carrier, let alone a known carrier, would be very rare: the recurrence risk of a Down's syndrome pregnancy after a previously affected pregnancy is approximately 1-2%.[185]

PGD for Down's syndrome has been reported in a few studies: in the case described by Cozzi and colleagues, at least 70% of ovulated oocytes were hyperhaploid for chromosome 21;[178] in the two cases reported by Conn and colleagues, 64% and 91% of unfertilised oocytes/embryos showed chromosome 21 aneuploidy and chromosome 21 imbalance respectively.[177] Though studies have demonstrated that the majority of second trisomy 21 pregnancies could be a product of chance alone,[186] cytogenic analysis can be used to rule out the possibility of parental translocations or mosaicisms[177] which may have implications for relevant risk of recurrence.[67;see 178] In those cases involving these predisposing factors, prospective parents who wish to avoid a child with Down's syndrome could opt for either: i) PGD; or ii) PND and (if desired) elective termination. PND for Down's syndrome is available through either chorionic villus sampling (usually conducted at 11-14 weeks of gestation) or with amniocentesis (performed after 15 weeks). Both methods involve an increased risk of miscarriage of approximately 1-2%.[187] The tests are invasive and carry slight risks to the foetus. As such, some reproductive clients find this route unsatisfactory owing to concerns about: i) the safety of diagnostic testing during pregnancy; ii) the risks of miscarriage; and, iii) ethical issues related to potential harms to the foetus.[187;188]

There are rare cases of women whose reproductive experience involves repeat pregnancies involving a conceptus affected by trisomy 21.[178] Hudson and colleagues reported on a woman who after two years of experience with infertility enlisted IVF services and successfully delivered a trisomy 21 infant. The patient then spontaneously conceived another infant with Down's syndrome, after which the woman and her partner had their supernumerary embryos (from the initial IVF cycle) genetically tested and screened for trisomy 21. Reportedly, "this couple did not consider a voluntary interruption of pregnancy a viable option in their circumstance."[189] For this couple—as for others who have the experience of living through the processes of repeat prenatal testing followed by elective termination(s)—PGD presented as an acceptable alternative to PND.[67;177;178] Thus, factor (a2) takes into consideration why certain couples who wish to reproduce might want to enlist PGD treatment services to prevent (or substantially lower the risk of) having a (or another) child with Down's syndrome (or perhaps some other genetic condition)[xxxi] as opposed to other alternative methods for doing so.

Factor (a1) speaks to the prospective parents' subjective views on the genetic condition to be avoided. To repeat: the HFEA guidance maintains that the only conditions for which PGD should be made available are those "where there is a significant risk of a serious genetic condition."[121:s.10.6] Though (as stated above) no official definition of serious has informed the law or policy, a public consultation document from the Authority explains that:

> How serious a condition is depends on how having the condition affects, threatens or limits the life of the individual, although these factors may be difficult to predict before the affected person is born. If the condition did not cause someone to suffer or detrimentally affect their life, the condition is unlikely to be regarded as serious. If, on the other hand, the condition required regular invasive treatment, or was life-limiting or life-threatening, it would be considered serious.[147:s.4.3]

Requests for PGD come almost exclusively from persons who have either first-hand experience of the toll of heritable genetic diseases for themselves (or sometimes their relatives) or of what it is like to live with the risks of being an identified carrier. These persons may also have experience of what it is like to witness the impact the condition in question has on siblings or their own children. Factor (a1) registers the HFEA's concern to acknowledge that the impact of being a carrier of a given genetic condition "can differ both in terms of how an individual might perceive the risk as well as, more practically, the way that the condition will manifest in any particular family."[147] As such, the HFEA has acknowledged that "people seeking treatment are in many ways best placed to judge the seriousness of the condition."[147:s.4.4]

[xxxi] There is of course the possibility that a couple's reproductive experience could include having already had a child with some form of a heritable genetic condition, and that they might want to now avoid not only the risk of this with a new pregnancy but also the risk of Down's syndrome. This would count as an instance of preimplantation genetic screening (PGS) which, though related to PGD, is outside the scope of this thesis.

In general, couples at risk of transmitting genetic conditions to their progeny recurrently experience stress, depression, and anxiety.[141] Mothers initially often feel guilty about having a child with Down's syndrome and, as is the case for many women, they are blamed by others and/or blame themselves whenever the results of reproduction do not meet with societal norms and expectations.[190] Unjust as this is, one study shows that (in time) mothers to children living with Down's syndrome score comparably to parents of non-disabled children in terms of levels of anxiety, feelings of guilt, and emotional burden concerning questions of responsibility for their children being the way that they are.[191]

The emotional and economic investment required for raising a child with a disability are not negligible (in the UK and most, if not all, other countries) and have been well documented in the literature. Certain individuals, couples, or families may lack access to needed resources, feel that they do not have the psychological wherewithal to raise a child (or another child) with Down's syndrome, or just prefer not to attempt such an endeavour. In these cases, where from the clients' viewpoint Down's syndrome presents as "serious", PGD treatment services may very well relieve anxiety and offer reassurance[65;see 141] at the earliest possible time—reassurance that the prospective offspring, in all probability, will be unaffected by trisomy 21.[xxxii]

The question is whether and, if so, how PGD is an appropriate "treatment" for addressing these sorts of needs. The HFE Act authorizes PGD as a treatment service in order to determine "that embryos are in a suitable condition to be placed in a woman,"[144:Sch. 2, s1(1)(d)] where "'treatment services' means medical, surgical, or obstetric services provided to the public or a section of the public for the purposes of assisting women to carry children."[144:s2(1)] In this regard, the term "suitable" has been given by the courts both a narrow[xxxiii] and a wide[xxxiv] interpretation in the case of *R. Quintavalle v HFEA*. On the narrow interpretation (as discussed by Lord Brown) PGD would be allowable only insofar as it is "necessary or desirable"[121:Sch. 2, s1(3)]

[xxxii] By speaking of reassurance and relief of anxiety that PGD prospectively offers some couples, this is not to say that it offers "mere" reassurance or relief of "mere" anxiety. Having PGD as a reproductive option has, in some cases, been all important to certain couples' decisions not just in how to reproduce but to reproduce at all.[52;192] As such, this technology has reportedly empowered certain women or couples at risk of transmitting genetic conditions and/or chromosomal abnormalities to take a "positive approach" to reproduction[193] and to advance their sense of reproductive autonomy.

[xxxiii] *Quintavalle v HFEA* [2002] EWHC 2785 (Admin), para.17.

[xxxiv] *Quintavalle v HFEA* [2002] EWCA Civ. 667, paras.37-45; *Quintavalle v HFEA* [2005] UKHL 28, paras.14, 24-28, 35, 56, 62.

for screening embryos to determine those that are *medically*[xxxv] suitable "to ensure that the woman can carry the child successfully to term—in other words embryonic screening to eliminate just such genetic defects as may affect the viability of the foetus and no other."[xxxvi] There is some evidence that embryos with numerical chromosomal anomalies, as with trisomy 21, have been significantly correlated with a higher incidence of implantation failures and repeat pregnancy loss.[194] It might seem, then, that embryo screening for these purposes could be justified on the narrow (medical) interpretation, where "suitable" is taken to mean those that have a better chance at initiating a pregnancy that will result in a live birth. At this point, the literature is inconclusive as to whether PGD in practice either reduces repeat pregnancy loss or IVF failure.[195] However, these findings are mainly based on the results of applying PGD for screening purposes for "all-comers to IVF" where the population is too varied, and has a low risk of relevant genetic "abnormalities", so any effect that one would see with PGD is diluted significantly. If one were to take only those patients with repeat pregnancy loss who have an identified parental translocation and utilize PGD to screen their embryos, certain experts in the field do maintain that this would probably increase the resultant live birth rate significantly,[196] so the same should be true for those women who are likely to be Down's syndrome carriers.

There is another possibility by which, on the narrow (medical) reading of "suitable", PGD could be used as a treatment for the mother's prospective anxiety at having a child (or another child) with Down's syndrome. But, it is to be remembered, that the courts' narrow interpretation (as discussed by Lord Brown) would only justify PGD screening of embryos for the purposes ensuring that "the woman can carry the child successfully to term": i.e., assisting the mother's health needs by way of PGD services is not in itself justified except in cases where it is undertaken to ensure the viability of the foetus. Within these parameters for justification, the use of PGD for trisomy 21 could be justified as a treatment service against the mother's anxiety of having a child (or another child) with Down's syndrome, but only if a causal link were to be established between the anxiety of the mother and the viability of the foetus. There is some evidence for this connection,[197] though some human reproduction experts are

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[xxxv] The distinction between medical versus social reasons for enlisting PGD, as well as the ethical and legal justifiability of each, is discussed in *Quintavalle v HFEA* [2002] EWCA Civ 667 at para.22: "Mr Pannick accepted that under this reasoning PGD with the object of ensuring that a child had certain characteristics for purely social reasons might also be said to be 'for the purpose of assisting women to carry children' but submitted that it was for the Authority to control PGD to ensure that this was not used for purposes which were ethically objectionable. That accorded with the scheme of the Act. It was only practices that were unquestionably objectionable that were prohibited by the legislation. PGD for the purpose of avoiding genetic defects was not objectionable at all."

[xxxvi] *Quintavalle v HFEA* [2005] UKHL 28, para.49.

sceptical.[198;199] In sum, the evidence is mixed to support PGD on the narrow (medical) interpretation as an effective treatment service for anxiety related to repeat pregnancy loss (in this case, for (likely) Down's syndrome carriers).

One can also read "into the statutory purpose specified by section 2(1)" of the HFE Act, "that of 'assisting women to carry children', the notion of healthy children—only a genetically healthy embryo being 'suitable' for placing in the woman within the meaning of paragraph 1(1)(d)."[xxxvii] On this wide interpretation of suitability, PGD is allowable as that which provides information about the make-up of the embryo and this information is relevant in providing assistance to the mother for deciding whether or not to endeavour to carry what would be the resulting child.[xxxviii] As Sally Sheldon comments: "The embryo to be implanted must be judged 'suitable' not just in narrow medical terms (in its ability to develop into a healthy child) but also taking account of the wishes of the mother."[200:408][xxxix] From this perspective, a woman's (or couple's) anxieties regarding what it would mean to have a child with trisomy 21 and the perception of what kind of impact doing so might have on the family could be deeply relevant as a factor in considering the appropriateness of PGD for a case of Down's syndrome. But it is important to notice that the evidence taken by way of factor (a1) here mostly points at ways in which PGD for Down's syndrome might be seen as an appropriate treatment alternative, but only as referenced against the particular wishes, subjective views, personal beliefs, and preferences of the mother or the parents together.

One counter-argument might be that for couples wishing to have a genetically-related child free of an inheritable genetic condition, PGD counts as an alternative which allows for testing at the earliest stage possible and obviates the need for later PND testing and the possibility (if desired) of elective termination. PGD has proven to be a preferred alternative in this regard insofar as it avoids the physical harm and emotional trauma that are associated with some women's experiences of abortion.[33;141;201;202] But, the risks and harms experienced through elective termination that are avoided by opting for PGD are not straightforwardly greater—indeed, they may very well be less so for some women—than the costs, harms, and risks (including in the UK a live birth rate of only 29.9% per cycle started[203]) involved with IVF[xl]—IVF

[xxxvii] *Quintavalle v HFEA* [2005] UKHL 28, para.51.

[xxxviii] *Quintavalle v HFEA* [2005] UKHL 28, paras.49, 60 and 62.

[xxxix] See *Quintavalle v HFEA* [2002] EWCA Civ. 667, para.128.

[xl] Having consulted with an obstetrician and gynaecologist (Dr. Robyn MacQuarrie[204]), I am told that from a medical perspective, the risks and harms to the mother as associated with IVF/PGD are definitely greater than those associated with PND/elective termination. Moreover, it should be noted that there are mixed reviews on how grave/onerous the experience is of having an abortion due to foetal anomaly. See

being a necessary step in PGD testing.[see 206;207] What is more, most clinics that offer PGD recommend confirmation testing later on with PND,[4;208;209] and so this step of prenatal testing with all of its risks and harms is still part of the PGD equation. Again, whether or not PGD is an "appropriate" treatment service for Downs' syndrome would seem to be a highly subjective determination and quite contingent on the client's preferences.

Effective therapy

"(c) the availability of effective therapy, now and in the future"[121:s.10.7]

Assessing the relevant evidence with respect to factor (c) is difficult since the term "effective therapy" is used without a definition in The Code of Practice (8th edition). Interestingly, the Joint Working Party of the HFEA and the Human Genetics Commission (HGC) originally recommended a more inclusive wording, stating that: "the availability of effective therapy *or management* now and in the future [emphasis added]" be taken into account when determining the appropriateness of PGD for any given condition.[29s.37,Rec.15] Down's syndrome is a chronic condition that presents from birth. Because there is neither a known cure for it, nor are there any probable, prospective cures in sight at this time,[cf. 210] most treatments aim to either control or mitigate adverse symptoms and to *manage* co-morbidities. It cannot be ignored that there are distinct medical challenges that come with Down's syndrome. For some parents, the health status of having a child with Down's syndrome can be a source of uncertainty, most notably in the first years of an affected child's life.[211]

The health conditions faced by persons with Down's syndrome can include: cardiac birth defects; congenital heart disease; hearing problems; gastrointestinal blockages; celiac disease; eye problems (*e.g.* cataracts); hypothyroidism; skeletal problems (*e.g.* hip dislocation); dementia; and a higher risk for acute lymphocytic leukaemia.[212;213] Interestingly, a longitudinal study by Carr that compared health at ages 4, 11, and 21 of persons with Down's syndrome with matched controls concluded that:

> the Down's syndrome group were not more often ill than were the controls, although, as Turner *et al.*[214] also show, illnesses tended to be more serious, especially in the profoundly disabled group. A small number suffered from multiple health problems and very poor health; nevertheless, compared with

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testimonials on "shared D & E experiences" at BabyCenter.com.[205] I underscore this point only to show that PGD can only be recommended as a suitable treatment service on the wider definition of "suitability" that references the judgments of the client/patient.

> the controls they did not as a group make excessive demands on family or hospital doctors. Most of those with Down's syndrome, like the controls, maintained good health over time, and in both groups poor health as a child did not necessarily presage poor health in adulthood.[215:115]

A recent, comprehensive review by Roizen and Patterson has shown that effective medical management of the health conditions associated with Down's syndrome is possible through organized assessment, monitoring, prevention, and vigilance.[216]

A major indication of improved quality of life for persons with Down's syndrome has been dramatic gains in the average life expectancy over the course of the past century, as accompanied by better overall health. A study that surveyed the survival trends from 1942-1997 of infants with Down's syndrome in "developed" countries (including the UK) observed an 87.8% life expectancy improvement.[217] Whereas in 1910 average life expectancy for this population in the UK was about nine years,[218] today it is between 50-60 years.[219;cf. 220]

Many of these gains in life expectancy are thought to be due to advances in knowledgeable medical care (*e.g.* cardiac surgery) and in general health management,[219] but other factors point to changes in terms of the social determinants of health (including mental health[221]) for persons with Down's syndrome.[222] A study that analyzed (by racial grouping) the death certificates of Down's syndrome Americans, found the average median age at death in 1997 to be 50 years for Caucasians, 25 years for African Americans, and 11 years for "other" races (including Asians, Hispanics, and First Nations Americans).[222] Friedman (the chief investigator on this study) noted two factors that could explain the "limited improvement" in median age at death for the American racial minority groups in this study: i) the frequency of life-threatening malformations; and, ii) differences in social factors and care[223] provided.[224] According to Friedman, there is no evidence that those persons with Down's syndrome from the indicated racial minorities were at greater risk of life-threatening malformations.[224] He concluded, saying:

> Because differences in ascertainment or severity probably do not explain these observations, differences in care received by persons with Down's syndrome might explain racial disparity in survival. Possibilities include differences in factors that may be associated with improved health in the general population such as socioeconomic status, education, community support, medical or surgical treatment of serious complications, or access to, use of, or quality of

> preventative health care. A combination of factors seems likely, as appears to be the case for racial disparity in mortality in the U.S. population in general.[224:465]

Other reasons explaining limited access to medical therapies for persons with Down's syndrome are of course diverse, but some may stem from structural components that work to systematically disadvantage this population. S.A. Cooper and colleagues [225;see 226] have aptly noted that people with intellectual disabilities—and this would include persons living with Down's syndrome[227]—experience health inequalities as compared with the general population: they have higher health needs levels[228-231] which are often not recognized and not met.[228;232;233] Access to healthcare services is also a greater challenge for persons with intellectual disabilities[234] (including persons living with Down's syndrome[235]) on account of: i) the learning and communication challenges of persons with disabilities; and, ii) beliefs and attitudes of healthcare providers.[225;236] Insofar as persons with Down's syndrome, as with persons with intellectual disabilities more generally, have different patterns of health needs, current public health strategies based upon the pattern of health needs of the general population can actually compound the health inequality gap.[xli;225;242]

In sum, persons with Down's syndrome are at increased risk for certain health problems as compared with the general population. In most cases these health problems are treatable with advances of modern medicine and increased access to medical care (when it is provided[see 243]). It would seem then that the evidence given in the literature shows that "effective therapies are available now"[121:s.10.7] for Down's syndrome.[244] Questions of access[243] to these therapies and their availability in the future, while posing challenges not to be under-estimated, obviously are

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[xli] See National Disability Authority (Ireland).[237] Cooper and colleagues have also pointed to the fact that persons with intellectual disabilities are discriminated against by way of participation in research. For instance, the Adults with Incapacity (Scotland) Act 2000 disallows those persons who are incapable of consent from participating in research unless the consent of their nearest relative or welfare guardian is obtained.[225] Those persons with Down's syndrome who fall under this category would therefore be prohibited from participation in research. Now admittedly—and with good reason—in cases where consent may be difficult or not possible to obtain from persons living with Down's syndrome, protectionist policies like this may be warranted and have been legitimately applied to vulnerable populations.[238;239] At the same time, the risks of avoiding research involving vulnerable persons also means that therapeutic interventions for these populations—when they are found, or when they are attempted, sometimes by necessity—are also riskier because they are either untested or inadequately tested.[240] The net effect of avoiding research for persons with intellectual disabilities, will also mean cutting off said populations, including in this case persons with Down's syndrome, from the prospective benefits of research.[241]

matters that *can be* addressed by way of social reforms [244], political will, and economic commitment.[xlii]

Intellectual impairments

(e) "the extent of any intellectual impairment"[121:s.10.7]

Determining the relevant evidence that applies in the case of factor (e) is challenging since nowhere in the Code of Practice is "intellectual impairment" defined. Clinically speaking, Down's syndrome is classified as a form of "mental retardation", though the term "learning disability" is now recommended in its stead in the UK.[see 246;247] Even so, little has been done to entrench this new terminology in UK law and references to "retardation"[xliii] are still standard in the medical literature. In this regard, the World Health Organization definition is commonly referenced which establishes mental retardation as a "condition of arrested or incomplete development of the mind, which is especially characterized by impairment of skills manifested during the developmental period, skills which contribute to the overall level of intelligence, i.e. cognitive, language, motor, and social abilities."[250:Ch.V (F70-F79)] Degrees of mental retardation are conventionally measured according to standardized intelligence tests. Individuals who test consistently below the intelligence quotient (IQ) level of (usually) 70 are classified as retarded.[250;251] Persons with Down's syndrome are expected to have moderate-to-severe mental retardation, with an IQ of 20 to 85 (mean score, approximately 50).[252] This means that 50% or more of this population function in the mild (IQ 50-69) or moderate (IQ 35-49) range of intellectual disability. Some have just a borderline degree of intellectual disability, while a minority have severe (IQ 20-25 to 35-40) intellectual disability.[190:117]

The average estimated intelligence of persons with Down's syndrome has been steadily increasing since the early 20th century.[253] At that time, persons with Down's syndrome were initially regarded as profoundly mentally retarded (*i.e.* IQ < 20).[253:35-37] According to Clunies-Ross, combined surveys of Down's syndrome children and adults from the first half of the century showed notable, widespread gains with most being classified as severely mentally retarded (IQ 20-34).[254] By the 1960s, reports estimated that 10% of cases were regarded as

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[xlii] In the United States, there has been a noteworthy drop in funding of Down's syndrome research at the National Institutes of Health, which decreased from $23 million in fiscal year 2003 to $17 million in 2008.[245].

[xliii] For a discussion of the controversy concerning the relationship of mental retardation to learning disabilities, see Siegel[248] and dell Orto and Power.[249]

mildly retarded; by the mid-1970s[xliv;254] these estimates increased to the point that researchers were suggesting that as many as 30-50% of older Down's syndrome children and adults were within the mild range for mental retardation, and even a few were thought to be within the normal range for intelligence.[256] Borthwick, whose study analyzes the above trends, observes that in the space of only 60 years, "[t]hese developments represent a general shift in the Down's syndrome mean of some 30 IQ points, from an average IQ of approximately 15-20 to an average IQ of, depending on the study, between 40 and 60."[257:407]

These findings are in some tension with: i) a common stereotype that individuals with Down's syndrome are "unintelligent, incapacitated and incapable of social function"[258;cf. 259] and, ii) the perception of mental retardation as "a permanent, unmodifiable trait attributable almost exclusively to physical causes"[260:314]—a prejudice that has been shown to foster community reactions of hopelessness, helplessness, and rejection.[261] There are several reasons to explain the stigma concerning the developmental potential of persons with Down's syndrome. Until the 1960s, effective education and training were rarely provided to persons with Down's syndrome, in part owing to the prevalent view then—and all too commonly still with us—that intelligence is a natural endowment, genetically determined and immutable, thus distinguishing those with mental retardation in a fundamental sense from the rest of the population.[257] As Clunies-Ross surmises: "In this climate, institutionalization seemed an appropriate alternative, but unfortunately the clients became a self-fulfilling prophecy. Little was expected of them, so they were taught little and therefore achieved little."[254:167] Only once the belief in "fixed intelligences" came to be challenged, did researchers[262;263] come to view experience and environment as having a major impact on cognitive development in infants and children.[254] Rynders and colleagues, after an extensive literature review (at the time in the 1970s), concluded that:

> For many years, the educational capabilities of Down's syndrome persons have been underestimated because a large number of studies purporting to give an accurate picture of Down's syndrome persons' developmental capabilities have had serious methodological flaws. A close examination of that literature, coupled with a look at findings from two current early education projects for Down's syndrome children, revealed that psychometrically defined educability

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[xliv] Clunies-Ross[254] points to a hallmark review by B.F. Kirman[255] that suggested that most children with Down's syndrome would classify as moderately (IQ 35-49) to severely (IQ 20-34) retarded, and an estimated 2-3% as mildly retarded (IQ 50-69).

> is far more common in Down's syndrome persons than a cursory review of the literature would lead [one] to believe.[xlv]

The review further revealed that researchers persistently assumed the false view that persons with Down's syndrome are all similar in terms of their capacities and developmental potential. They therefore overlooked individual differences in performance and ability across this population.[254]

For some time now it has been widely agreed that the developmental characteristics of Down's syndrome children are extremely variable[xlvi] and that motor, mental, and social development is faster for those raised at home than in institutions.[xlvii] Francis further established that adverse behavioural changes in older children with Down's syndrome were on account of institutionalization and not, as previously thought at the time, because of the effects of aging.[xlviii] Connolly (among others[xlix]) in a study involving non-institutionalized children with Down's syndrome, has also observed the significant advantaging effects of early intervention programming to enhance intellectual and adaptive functioning in these children as compared to age matched controls.[l] Clunies-Ross concurs with these findings by observing that:

> Criterion-referenced assessments of older children involved in the EPIC [*Education Program for Infants and Children*] and similar programmes[273;274] have demonstrated that it is quite realistic to expect many (and probably the majority of) 5-6 year old children with Down's syndrome to be able to perform well over 90% of the tasks expected of non-disabled children of the equivalent CA [chronological age].[254:168]

With regards to further accounts in the literature on relevant intellectual impairments, children with Down's syndrome are known to face particular challenges in verbal—though not necessarily communication [275:424]—and academic skills, especially those related to arithmetic and reading.[253;276] Even so, Buckley has noted that they can progress to levels of academic skill[li]

[xlv] Rynders[264:440] quoted in Clunies-Ross.[254] See also Wallin.[265]

[xlvi] Levinson[266] cited in Clunies-Ross.[254]

[xlvii] Centerwall,[267] Melyn,[268] Stedman,[269] and Shipe[270] cited in Clunies-Ross.[254]

[xlviii] Francis[271] cited in Clunies-Ross.[254]

[xlix] For examples see citations 10-17 of Clunies-Ross.[254:167]

[l] Connolly[272] cited in Clunies-Ross.[254]

[li] One case study of a research participant with mosaicism of trisomy 21 reports that he demonstrated normal intellectual development up to the point of achieving "good" academic progress at the university level.[277] L.M. de A Moreira and colleagues further concluded that, along with other possible genetic determinants, "strong family support, early and continued intervention programmes for both physical and speech therapy, and a thorough educational process, also provided opportunities for the

that have surpassed researcher predictions.[278] In the late 1980s, Carr conducted a longitudinal study that involved Down's syndrome research participants (aged 6 weeks to 21 years old) as well as their families. Carr found that "where verbal and academic skills are concerned there appear to be clear environmental effects, with the young people from more [socio-economically] advantaged backgrounds doing significantly better than those from the less advantaged [backgrounds]."[275:424] Carr's study did not control for how much deliberate teaching was imparted by the parents of the "advantaged" children, but her anecdotal impressions lead her to surmise that: "the influences at work seem likely to be the more subtle ones of atmosphere and expectations, of availability of relevant materials—books, newspapers, magazines—of modelling of skills by other family members and of the opportunity to join in with skill-related activities."[275:425] [lii]

The intellectual impairments of persons with Down's syndrome may also be overstated due to confounding co-morbidities or other limiting physical impairments.[246:815] Turner and colleagues have noted that up to 77% of this population have some visual impairments and 62% experience hearing loss.[214] At the same time, cognitive functioning and learning disabilities are known to be masked by such physical impairments,[281] and very little work has been done to sort out the effects of this for assessing the intellectual capacities of persons with Down's syndrome.[257:407-408] Libb and colleagues have observed that children with Down's syndrome who had abnormal tympanograms (which might imply middle ear disease) performed less successfully on intelligence tests than did those children with Down's syndrome who had normal tympanograms.[282]

There is some evidence that this population experience more rapid cognitive deterioration than typically developing persons.[283-285] [286;cf. 287] But as Carr cautions: "...it is unsafe to attempt dogmatic prediction about the future developmental level of any individual with Down's syndrome."[275:423][liii] Gillberg and Soderstrom[246] in one study, and Burt and colleagues in

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development of the cognitive potential of the subject."[277:91] This said, mosaic trisomy 21 is not typical trisomy 21, and these individuals generally do better intellectually.

[lii] Dykens has also suggested that having sustained and structured stimulation are protective factors that can buffer against the effects of psychopathology in persons with Down’s syndrome. Dykens states that "[e]xamining youth with Down’s syndrome, Shepperdson[279] found that higher levels of environmental stimulation from school, social, or other daily activities were the single best predictor of successful functioning in adult life."[280:276]

[liii] Cooper has pointed out that elderly people with learning disabilities (which would include persons with Down's syndrome) in England have greater psychiatric morbidity too. However, in this study, elderly people received less day and respite care, availability of a social worker and most health services than younger controls. Cooper has noted that: "Failure to access services may relate to carers' attitudes and beliefs: in learning disability settings, morbidity was attributed to 'it's just old age'; in the elderly

another,[290] found that Down's syndrome research participants with increased IQ tended to experience less cognitive deterioration than matched controls with lower IQ scores. Given that IQ may be associated with level of education, Gillberg and Soderstrom hypothesized that improving cognitive functioning could be useful for deferring the onset of dementia.[see 291] Their study also associated "good caregiver support …with a slower decline in cognitive abilities in Down's syndrome."[246:818] Oliver and colleagues further established that: "When age was controlled for, cognitive deterioration was significantly positively associated with caregiver difficulties and service use and negatively associated with life experiences for the individual. Results suggest a potential role for caregiver difficulties in influencing life experiences of adults with Down's syndrome showing cognitive decline."[292:455] Hence it is evident that various confounding, contingent, situational, or environmental factors—many of which can be corrected for, according to a growing body of evidence[liv]—are implicated in the overall course of intellectual development and cognitive capacities of persons with Down's syndrome.

The effects of intellectual impairments for persons with Down's syndrome have thus been exaggerated through history, and the differences in cognitive functioning as measured by IQ[lv]

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settings, morbidity was attributed to 'it's because s/he has learning disabilities'."[288:331] See also Cooper.[289]

[liv] Buckley notes that: "A small number of studies have looked at the progress of children with Down syndrome in relation to family characteristics and coping strategies. Like all children, the progress of children with Down syndrome is influenced by their family environment, and their educational and social opportunities, not just determined by their genetic makeup. Children with Down syndrome show the usual benefits of social class, with those in families with more resources and higher levels of parental education making better progress. They also show the same benefits of family position, with first and only children making better progress, presumably due to more individual adult attention.[293] ¶In the Manchester studies,[294-297] the findings indicate that progress in self-sufficiency (life skills) and practical skills were more influenced by family factors than academic progress. Higher self-sufficiency scores were achieved by children whose mothers used practical problem solving strategies and lower for those who tended to use wishful thinking, even after taking account of the effects of mental age on the children's progress. As the children got older, the link between cognitive ability and life skills became weaker. Cognitive ability as measured by mental age predicted academic achievement but family factors and social opportunities had more influence on life skills."[298:n.p.(online)]

[lv] Thus far my argument has assumed the credibility of IQ measures. One could also argue that not only have persons with Down's syndrome been misjudged by IQ measures, but that the instruments employed to measure IQ are inadequate to gauge intelligence for a variety of reasons. For instance, critics have objected that IQ scores tend to measure a very narrow band of intelligence types, with often a heightened emphasis on analytic skills.[299] Feminists[300] and other critics have been instructive here for making plain that IQ overlooks emotional and other kinds of intelligence.[301-303] As such, persons with Down's syndrome are reputed for strengths in nonverbal social interaction skills,[304;305] distinct and sometimes heightened powers of empathy,[306;307] as well as general social competence skills,[308] which are not the kinds of cognitive capacities that figure on IQ tests[309:63] nor in cultures that stress individual competition, but which are no less real or intrinsically valuable on that account.[310] The claim here should not be overstated. Recent studies show that persons with Down's syndrome, though adept at facial recognition, experience challenges in recognizing differences in emotions as portrayed through facial expressions.[311]

can be accounted for (at least) in part by social deficiencies unrelated to "what the standard classification describes as 'significantly subaverage general intellectual functioning.' "[257:404] Borthwick further argues that just as racial minorities have been unfairly disadvantaged on IQ testing, so also prejudice against persons with Down's syndrome is likely to unfairly disadvantage this population on IQ testing with the effect of yielding inaccurate and spuriously depressed scores.[257403-410] He suggests that "people with Down's syndrome may be, precisely, handicapped, as racehorses are handicapped—that they carry lead in their saddlebags from the effects of prejudice and physical disability, and if it was possible to remove these then they would be expected to come further up the list of finishers than they do."[257:407-408]

In all likelihood then, some of the developmental gains and greater ability to overcome impairments in the Down's syndrome population (as noted above) are probably owing to early intervention strategies[312:108-109] and changes to educational programming.[313] Some of these notable changes to educational programming have involved shifting away from the custodial, management model of care set in the context of segregated institutions[253:35-37] to person-centred models of care and education (which aim at broader goals of social inclusion[314]), giving concerted attention to the specific learning styles and motivational factors of *individuals* living with Down's syndrome.[311;313;see 315] Although persons with Down's syndrome face undeniable intellectual challenges on account of having certain intellectual impairments, the above evidence shows that the source and degree of the consequent experience of disability is contingent upon several social and environmental variables—variables that can be addressed to substantially obviate, or at least mitigate, the potential for disability associated with Down's syndrome.

Associated suffering from the perspective of the individual

> "(b) the likely degree of suffering associated with the condition"[121:s.10.7]

The expected suffering associated with a given genetic condition is likely one of the factors most weighted in decisions considering the appropriateness of PGD. On the face of it, it makes sense from a parent's perspective, that if given the choice with PGD, it is preferable not to bring into the world lives that are very likely to be dominated by severe and chronic suffering.[175;316] I have argued elsewhere[175:190] that it is often the incurable, intractable pain and resultant severe

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Wishart concludes that "[t]hese findings are consistent with the emerging picture of neurological disruption in Down syndrome and with what is known of the neurology underlying this key component in social cognition."[311:47] suffering associated with certain genetic conditions that might preclude a person from having the opportunity "to achieve a decent minimum level of lifetime well-being",[317:199;318;319] or quality of life that, taken together, might not make for a "decent chance of a reasonable life",[17;320:670] or (in the extreme) "a life [perhaps] not worth living."[321:16] All lives assuredly will involve some suffering.[75:99] The question before us is whether a life with Down's syndrome involves risk of undue suffering. I look at this question first by assessing the relevant evidence as given from the perspective of the individual, and then, in the next section, from the family's perspective of living and supporting a family member with Down's syndrome.

At this time, it would appear that there is no large study on how people living with Down's syndrome judge the quality of their own lives,[322;323] though some smaller qualitative studies have been conducted involving interviews with individuals from this population.[221;324] Anecdotal reports[lvi] confirm that these persons do not consider having the condition (itself) to be a source of suffering, [322;324;327] nor that their lives on this account are seriously limited,[328:i] onerous, or "handicap[ped]".[see 326;327:35] Instead, amongst relevant interviewees and advocates a prevalent impression voiced is that society's behaviour in response to them is seen as a primary source of what suffering there is in their lives in the forms of prejudice and social discrimination.[324;329] As Anya Souza, a writer with Down's syndrome, reports: "My message is that we can achieve a lot more than people imagine, and we deserve more respect. ...I don't feel sorry for myself and I don't want anyone else to feel sorry for me, either. I enjoy my life so there's no need."[330] Marianna Paez, an advocate with Down's syndrome adds: "... it doesn't matter to have Down's syndrome. It matters to be alive and to feel and hope like everyone else."[331:60]

It might be objected that the first-person accounts in these studies and testimonials come from *select* individuals who are—as some researchers describe—"high functioning" and therefore not representative of those that face more profound lack of facility with language and communication barriers.[lvii] Admittedly, personal life satisfaction assessments for nonverbal

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[lvi] As Scott[325] has noted, qualitative researchers investigating the self-perception of persons with Down's syndrome, like Priscilla Alderson,[326] acknowledge that this kind of research is not necessarily representative for the population. This points to a need for further research of this kind in this area. Again, without further research, persons with Down's syndrome may not be receiving the potential benefits that would come from such research, even if the benefits are indirect.

[lvii] Children with Down's syndrome also could be included here. Interestingly, Roizen and Patterson have observed that "[p]arents frequently report that the child with Down's syndrome is happier and more loveable than other children, although investigators have found children with Down's syndrome to be of similar temperament to other children."[216:1287] See Wishart[311] for an explanation of how these "positive" stereotypes can actually impede expectations and learning for children with Down's syndrome.

persons with Down's syndrome (as with nonverbal persons with disabilities in general) point to a lacunae in the research and literature, even though, as Alderson points out: "research through interactions and observations with people who do not speak about their quality of life [is possible and] can be very informative."[313;332;see 333] Skotko and colleagues from a survey of 284 persons with Down's syndrome (age ≥12) found that nearly 99% "indicated that they were happy with their lives, 97% liked who they are, and 96% liked how they look. Nearly 99% …expressed love for their families, and 97% liked their brothers and sisters."[334:2360] This is not to downplay the challenges (from a first person perspective) of a life with Down's syndrome,[280;335] but rather to point out that the experience of a life with Down's syndrome is not co-extensive with a life of suffering. More on point research is needed in this respect.[174;see 324]

It would seem, then, that from a first person perspective on the issues, suffering associated with Down's syndrome is to some extent exaggerated.[lviii] As certain disability activists have explained, a good life with Down's syndrome is very possible and much of the hardships endured are experienced as being on account of social prejudice and discrimination,[lix] though further research is needed to confirm these impressions.

Associated suffering from the perspective of the family; the impact of social supports and family circumstances of the people seeking treatment[lx]

> "(b) the likely degree of suffering associated with the condition"
> "(f) the social support available", and
> "(g) the family circumstances of the people seeking treatment."[121:s.10.7]

As stated in the introduction, over the past decade the HFEA has come to place more emphasis on families' experiences and their perceptions of the gravity of heritable genetic conditions in determining the appropriateness of PGD.[337:10;338] The suffering associated with Down's

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[lviii] Perspective matters here. See Scott for a discussion of how, in general, "people with a given illness or condition report greater quality of life compared to that attributed to them by others.[326]"[176:221-222] See also Janvier et al.[336] for a study of the markedly different interpretations between families and healthcare providers about the experiences of disabled children and their quality of life.

[lix] An important aspect of the social model of disability (which we will come to shortly) has been in drawing attention to the social sources of disability, including the experience of unfair discrimination and lack of social supports.

[lx] Interestingly, the "seriousness" of a prospective case for PGD is not determined in consultation with social workers or service providers—only between parents and medical personnel and perhaps genetic counsellors.

syndrome is therefore not only to be evaluated according to the interests of the "child/individual-to-be," but also according to the collateral effects—including perceived effects, as with factor (a1)—that bear on the interests of the whole family.[see 176:ch.1 & 244-245] In this regard, on a wider scale, the oftentimes presumed negative impact of disabilities on families' quality of life is coming under scholarly and political challenge[339:72-73;see 340] as unjustified in the face of a rising body of evidence showing the contrary from research on families with children with impairments.[336;341-348] More specifically, psychological profiles and functioning measures of families of children with Down's syndrome are reported as being (for the most part) very similar to their relevant, so-called "normal", counterparts.[215;349;350][lxi] I turn now to assess the specific evidence of the experience of families who live with, and care for, members who have Down's syndrome, and to highlight aspects of their circumstances and available supports that have a significant bearing on the varied character of this experience.

It is common for parents to experience a sense of disappointment if their foetus or newborn is diagnosed with Down's syndrome. Nevertheless most parents to infants with Down's syndrome become attached within a few months following birth.[216:1287] King and colleagues reported that while parents of Down's syndrome children grappled with "lost dreams", over time, through changing their belief systems concerning life, disability, and the family, most reported increased feelings of empowerment, personal growth, and rearrangements of priorities as instances of positive adaptations to parenting a child with a disability.[351] Van Riper conducted a study in 2007 that used the *Resiliency Model of Family Stress, Adjustment, and Adaptation* to assess maternal perceptions of parental and family adaptation in families of children with Down's syndrome. Seventy per cent of the seventy-six mothers polled reported their overall family functioning as either 4 or 5 on a 5-point scale (1=poor; 5=excellent).[352] Hodapp and colleagues found no significant difference between parents of Down's syndrome children and parents of typically developing children with respect to their perceptions of the rewards of parenting their children.[353] Cuskelly *et al.* actually found that "mothers of a child with Down syndrome reported significantly more reinforcing aspects to their relations with their children than did mothers to typically developing children."[312:107;see 354] With a "valid and reliable survey instrument", Skotko and colleagues in a high powered study that polled 4,924 households from the mailing list of six non-profit Down's syndrome organizations in the United

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[lxi] Ferguson *et al.* note that "there is a level of agreement approaching consensus that the overall adaptational profile of families who have children with disabilities basically resembles the overall profile for families in general (including children with and without disabilities)"[347:81] and also that "family responses to disability are immensely variable."[347:85]

States found that: "Of the 2,044 respondents, 99% reported that they love their son or daughter; 97% were proud of them; 79% felt their outlook on life was more positive because of them; 5% felt embarrassed by them; and 4% regretted having them."[355:2335]

Parents who raise children with Down's syndrome are often very aware of the prevalent and persistent effects that the child's unique needs can have on siblings.[356;357] In this regard, various studies have reported on parents who testify to the benefits of having a Down's syndrome child for their other children.[312;358] Children with Down's syndrome, for instance, manifest characteristics that have worked to maintain and develop relations between family members and "significant others" outside the family.[359] Lobato and colleagues investigated similarities and differences between young siblings (aged 36-78 months) of children with disabilities (including some with Down's syndrome) as matched with controls of siblings without disabilities. The study found few differences between the groups with respect to the quantity and quality of their interactions. In fact, siblings of children with disabilities engaged in more parallel and social play than did the matched controls. Siblings of children with disabilities in this study also showed themselves to be more nurturing "but no more likely to interact aggressively or to be commanding or directive with their brothers or sisters."[360]. Cuskelly and Gunn found that siblings of children with Down's syndrome "reported less unkindness and, if in a same-sex dyad, more empathy than did comparison children."[361:234] In a recent review article, Cuskelly and colleagues [312] concluded that: "findings suggest that there are no important differences in the adjustment of the siblings of a child with Down's syndrome and children in families where all are developing typically[349;362] and that relationships are as good as, or better, than in these families."[312:107;see 363] Skotko and colleagues, in the high powered study mentioned above, found that parents to children with Down's syndrome reported that "95% of their sons and daughters without [Down's syndrome] have good relationships with their siblings with [Down's syndrome]."[see 334;355:2335;364]

Research has also shown that (neurotypical) siblings to Down's syndrome relations have favourable self-concepts.[365] Carr found that these (neurotypical) siblings did not feel themselves unduly burdened by having a brother or sister with Down's syndrome.[275:427] Skotko and Levine, in a recent widespread study of siblings to persons with Down's syndrome, found that said siblings reported a wide range of emotions and that, on balance, the positive feelings were experienced as outweighing the negative ones. These researchers concluded that "siblings find rich value in having a family member with Down's syndrome, and most will assume positions of advocacy at some level in their lives."[see 310;358:186]

Although adverse effects are less than might be expected by many parents and siblings, there are definite challenges for families with one or more members who have Down's syndrome.[366] In a study on family welfare, Brown and colleagues observed that families with members who have a disability registered lower quality of life with respect to measures of health, social support, finances, and career opportunities as compared with families with only typically developing children.[367;see 368] Carr reports that especially mothers carry significant burdens with respect to poorer health, reports of more depression, and most significantly, their *Malaise Scale* scores were higher than matched controls.[275;369] Building from the work of Ferguson and Watt,[370] Carr also noted a significant relation between *Malaise Scale* scores and social class—an association that presented as more closely related than between *Malaise Scale* scores and the level of the Down syndrome child's disability. In previous studies, neither Bradshaw[371] nor Carr and colleagues[372] were able to significantly associate *Malaise Scores* with a number of objective factors, including the child's level of disability or measures of social restriction.[275:427] More recently, van Riper found a significant association between these factors and the relevant mothers' perceptions of the quality of support (which they and their children with Down's syndrome) received from the professionals that were charged to work with them.[373]

Gallagher and colleagues found that "caregiver burden, in general, and its guilt component, in particular, predicted symptoms of depression and anxiety in parents of children with intellectual disabilities," including parents of children with Down's syndrome.[374:1129] More generally, patterns of parental stress in families with members who have a child with a disability (also including those with Down's syndrome children) are reported to track gender distinctions.[345;369;375-378] Roach and colleagues have observed that parents of children with Down's syndrome perceived more care-giving difficulties, child-related stress, and parent-related stress than did matching controls of parents with typically developing children. While mothers' stress tracked experiences of difficulty related to responsibilities of caring, fathers' stress tracked experiences of difficulty related to accepting their Down's syndrome children's group status as "non-normal" or "disabled."[see 369;379] Those mothers that assumed disproportionately higher care-giving duties reported more difficulties with their own health, sense of autonomy (*i.e.* role restriction), and partner support. Conversely, fathers who assumed more responsibility for relevant childcare reported not only fewer difficulties with attachment but also a greater sense of parental competence. Spousal stress correlated with both mothers' and fathers' stress.[379] This study suggests that distribution of responsibility for care, if realigned to avoid gender stereotypes—where mothers assume disproportionately higher responsibilities for care of children[see 366]—can significantly impact the experience of stress associated with raising a child with Down's syndrome.

Hodapp has observed that most studies focussing on families' experiences of having a member with Down's syndrome, examine only parental or sibling levels of stress and coping strategies. Increased attention is needed to determine the effects of spousal, occupational, health, educational, and other socio-economic outcomes for family members.[380] In this regard, more generally, the prevalent impact of the social determinants of health is well known.[98;381] They are known to have an over-riding impact on: i) the health of children with intellectual disabilities;[382;382-384] ii) reported satisfaction of adults-with-intellectual-disabilities concerning their own level of well-being;[385;385;386] iii) family functioning of families with children with intellectual disabilities;[387] as well as (iv) the health and well-being of parents to children with intellectual disabilities.[388] Hodapp concludes the above-mentioned study, saying: "Although [Down's syndrome] families have received some research attention over the past several decades, we now need to make family research in Down's syndrome more concrete, more life-span, and more tied to characteristics of the individual with the syndrome and the family's surrounding support system."[380:279]

The parental experience of raising a child with Down's syndrome, as well as the experience of living as, and with, a Down's syndrome family member, is of course variable. As reported in the literature, family circumstances, the level of social supports that are made available to the relevant families,[221;see 389] and other social factors[369] can be reformed to either mitigate or obviate much of the relevant individuals' and families' experiences of suffering and/or compromises to their quality of life.[324;390] The latter point speaks directly to whether Down's syndrome ought to be considered a "serious" genetic condition, and what is meant by continuing to consider it a "serious" condition—matters which we shall focus on in the next section and also the next chapter.

Models of disability: locating the source of, and determining how best to respond to, functional limitations

The likelihood and expected extent of suffering associated with a given condition is taken as a most significant consideration in gauging whether or not PGD is an appropriate intervention so as to prevent the birth of relevantly affected children. Trying to prognosticate the likelihood and expected extent of suffering are related to how one understands the different kinds and different sources of disability associated with a given condition. Clarifying these relations is important for understanding and assessing what makes a condition considered "serious" or "not-serious", as well as the reasons prospective parents and their supporting AHR service providers might consider PGD as a way to avoid the risk of having a (or another) relevantly affected child. What presents, or (alternatively) is recognized, as a disability has a long history of dispute across disability theorists. Models of disability, as Scott aptly observes, not only affect our perception of disability; they also affect "the desirability of avoiding it."[325:66] In this section I propose to consider certain anticipated criticisms of my assessment of the source(s) of disability for persons living with Down's syndrome by engaging with pertinent debates in the literature (mostly from the UK) between the competing medical and social models of disability. In short, these models argue for differing accounts of the relationship between impairments and disabilities and sometimes disagree about the distinction between disability and (social) disadvantage.[lxii]

It is a reality that persons living with Down's syndrome face all sorts of disadvantages.[lxiii] Broadly speaking, many of these disadvantages can be seen as a product of their interaction with society as persons with functional limitations. Hull explains this relation, saying: "Without society's being as it is, people's functional limitations would be different, no less than society would be without people with functional limitations."[397:203] On a prevalent reading of the medical model of disability, disability marks a disadvantage stemming from impairment(s)—a restriction or lack of ability to perform "normal" human activities.[398:659;see 399:99] Briefly put, medical models of disability tend to locate the source of disability in what is understood as the inherent loss of functioning owing to physical or intellectual impairments in the individual. Medical models (in general) counsel the investigation and delivery of effective forms of medical

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[lxii] Here it is important to consider how disability and impairment are defined. This is contested conceptual territory with much disagreement, but nonetheless some international consensus. For instance, the World Health Organization (1980) defines *impairment* as "any loss or abnormality of psychological, physiological, or anatomical structure or function" and *disability* as "any restriction or lack (resulting from an impairment) of ability to perform an activity in the manner or within the range considered normal for a human being."[391] Although these definitions are still quite commonly referenced in the literature, they have been updated in *The International Classification of Functioning, Disability and Health* (ICF), which views *disability* and *functioning* as "outcomes of interactions between health conditions (diseases, disorders, and injuries) and contextual factors."[392:10] In turn, the ICF recognizes three levels of *human functioning*: "functioning at the level of body or body part, the whole person, and the whole person in a social context. Disability therefore involves dysfunctioning at one or more of these same levels: impairments, activity limitations and participation restrictions."[392:10] *Impairments* are defined for the ICF as "problems in body function or structure such as a significant deviation or loss."[392:10]

[lxiii] For relevant American studies see: National Organization on Disability (NOD) and Louis Harris & Associates,[393] NOD,[394] and Taylor.[395;396]

treatment for affected individuals' impairments to either manage symptoms associated with non-normal functioning or to assist the individual in regaining or, as the case may be, restoring them to normal functioning as much as possible. Medical models of disability also put a great deal of emphasis on disease/disorder prevention.[398:659;cf. 399:99;400:33]

On the other hand, social models of disability tend to emphasize a strong distinction between having impairments and being disabled,[401;402:92;403:358] such that functional limitation or impairment does not necessarily entail disadvantage.[397:204] Social models argue that the main sources of disability reside in the ways that society is structured to exclude, or insufficiently accommodate, the different needs of persons with impairments.[lxiv;400:34;402] Most theorists who propound a social model of disability maintain that whether an impairment results in a disability is contingent upon the character of the environment in which the individual lives. A person's social environment consists of both material-physical and symbolic-interactive dimensions.[69;404:xii;see: 405] Iris Marion Young gives the example of how moving on wheels presents as a "problem" or "disadvantage" for environments with stairs and other radically uneven surfaces. So also, in cultures where making eye contact is a normal expectation during conversation that counts as a sign of respect and evidence of "engagement", those who do not do so (for a variety of reasons) are commonly disliked or feared as a result. As Young summarizes:

> The social model of disability notices many such attributes of environments and relations among persons, and the project it has generated would systematically describe the structures and processes of such social environments as they construct some people as 'normal' and others as 'deviant', giving many advantages to the former.[108;404:xii-xiii;see 406]

[lxiv] This is sometimes also referred to as the "Minority-Group Model of Disability". Waddington explains: "This is a socio-political definition which argues that disability stems primarily from the failure of the social environment to adjust to the needs and aspirations of disabled people, rather than from the inability of disabled people to adapt to the environment. The argument here is that it is *discrimination,* in both the physical and attitudinal environment, prejudice, stigmatisation, segregation and general history of disadvantage, which we have come to associate with disability (but which need not be), which is the major problem for disabled people. According to this perspective the difficulties confronting disabled people come from the disabling environment rather than from within the individual. This can be seen as a fundamental reconceptualisation of disability and, since it focuses [on] deficiencies in the environment instead of those in the individual, as the functional-impairment model does, it has wholly different policy implications. If the problem lies in the environment and society, it is these elements which must adapt, until a world exists which is suitable for those who deviate from the expected physical and mental 'norms'."[400:34]

Accordingly, disability is commonly seen from this point of view as a form of social oppression that is institutionalized in societies; by contrast, impairment is seen from this point of view as "nothing more than a description of physical, sensory or mental difference."[403:358][lxv]

Proponents of a social model of the disability might want to argue that Down's syndrome is classified as a "serious" condition because of a misunderstanding of what life is like for the relevantly affected individuals and their families. Linked to this claim, is the question of whether the impairments that come with Down's syndrome do not have to be disabling, but are instead "one more form of 'neutral' human variation just as non-disabling traits like left-handedness, sex, [or] skin colour."[171:43]

But, critics of the social model of disability and this sort of assessment of Down's syndrome could rejoin that (surely) not all problems of functionality that characterize a life with Down's syndrome are due to inadequacies of social response.[325:67] They might argue that Down's syndrome is not labelled as a "disability" without (any) reason: after all, any resulting life with Down's syndrome is likely to be more difficult because of it.[408:127] So, even if a life with Down's syndrome does not make a fulfilled life impossible, it is not just one more form of "neutral" human variation such as the examples given above.[cf. 171:43] To think otherwise is to confuse disability with disadvantage. These same critics could make a strong case that the phenotype for Downs' syndrome is variable,[409;410] and that, at a minimum, certain expressions of the genotype are not completely benign or neutral, but would entail disability even in the most supportive, social environments.[lxvi] From this viewpoint, persons born with Down's syndrome risk losses that are different from being just socially disadvantaged.

In this regard, Sally Sheldon and Stephen Wilkinson[413], as well as others,[325:67;399] have worked to clarify the distinction between disadvantage and disability—two distinct notions

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[lxv] Reindal, a strong proponent of the social model of disability, points out that: "To deny that there is a necessary causal link between a certain condition in the individual and disablement does not imply that researchers defending a social model deny that there are such things as impairments. On the contrary, impairment simply means that aspects of a person's body do not function or they function with difficulty.[407:55] What they insist on is that whether an impairment results in disablement is conditional on other things than merely the impairment itself. They acknowledge the fact that individuals might regard their impairment as positive, neutral or negative, and that this might differ according to time and place. Hence, they favour a social model of disability where the basis of disability is located in social conditions. Advocates for the social model maintain that disability is a socio-political construction, a product of organisation and culture, rather than a personal limitation due to a person's impairment, where agency and structure are intricately knit together. Such a view implies that social barriers, norms and values, rather than impairments, should be analysed in determining quality of life."[402:92]

[lxvi] Baily notes the unknown variation in severity of a condition may well affect decision-making in people who are risk averse. Baily explains: "A woman's decision may reasonably be influenced more by the 'worst-case' Down syndrome scenario than by the average."[411:70] For further discussion of this point see Steinbock.[412]

which are sometimes run together in social models of disability.[325:67] Following Sheldon and Wilkinson's analysis then:

> [a] reason for rejecting the social model is that it ignores the obvious fact that some of the disadvantages associated with disability are not caused by society, but are intrinsic parts of the impairment itself.[413:103] ...Where present, these things are intrinsic 'evils': harms which are not caused by society and which, arguably, no amount of social intervention, short of removing the impairment, can take away. Social factors are not the whole story.[413:104][lxvii]

As such, Sheldon and Wilkinson appeal to a less polarizing discussion of disability,[lxviii] and so they point to the work of Richard Hull (mentioned above) who advances the view that there are impairment-induced disabilities as well as socially-induced disabilities.[lxix;397] As Hull states: "Any definition of disability, if it is to reflect accurately the *experience of disability*, must pay homage to both the reality of impairment and the reality of social discrimination [emphasis added]."[397:209] Even exponents of the social model of disability like Tom Shakespeare[lxx] have done much to clarify the limitations of (sometimes) extreme social constructionist interpretations[cf. 415] of disability[416] that problematically underplay the relevance of medicine[lxxi]

[lxvii] Sheldon and Wilkinson further explain that while the disadvantages faced by being a woman in patriarchal cultures are all (or almost completely) caused by social discrimination, only some of the disadvantages faced by disabled persons are caused by social discrimination.[413:104] They conclude, saying: "It is plausible to suppose that whilst all (or virtually all), of the special problems faced by women are caused by social discrimination, only some of the difficulties faced by disabled people are caused by such discrimination—the others being caused by impairment."[413:104-105] There is an involved literature between those who distinguish and those who dispute the difference between disadvantage and disability. For further clarification in these matters see Hull.[397]

[lxviii] This distinction is sometimes overdrawn. See Glover.[17] Tom Shakespeare warns of polarizing discussions of disability that entrench dichotomies of "medical versus social", "impairment versus disability", and "disabled versus non-disabled people".[75] Susan Wendell asserts that "[c]areful study of the lives of disabled people will reveal how artificial the line is that we draw between the biological and the social."[414:266] In this respect, Shakespeare's and Wendell's points track certain feminist concerns to resist dichotomous thinking which would underestimate, or disregard altogether, relational components of health and the values of interconnection and interdependence that underwrite the relevant practices.[114]

[lxix] Jonathan Glover echoes this point of view from Hull, saying: "There has been a debate over whether we should replace the 'medical' or 'functional' model of disability with the 'social-construction' model. It is time to give up this debate, as it is now unfruitful for the same reason that makes the 'nature-nurture' debate unfruitful. To many disabilities, there is a contribution from a variety of sources, including functional limitation *and* social context."[17:7-8]

[lxx] To be more accurate, Shakespeare has been throughout his career both an exponent and a critic of the social model of disability. See Shakespeare.[75:5-6]

[lxxi] As Shakespeare notes: "Despite the deficiencies of medical sociology and medical ethics, it would be foolish to ignore the many valuable contributions to understanding disability made from these perspectives. Moreover, it is not medicine, but inappropriate medicalization which is the root of the problem."[416:11]

for understanding (and redressing) how impairments can circumscribe functionality and agency.[lxxii;75] Shakespeare owns that some impairments, in and of themselves, present significant problems as well as "broader cultural and political consequences."[416:13] His objection to the social model is that "it fails to capture the complexity of disabled people's lives."[416:11] Rather than treating all disability as one class—either medical, or social—Shakespeare suggests that "[i]t is time to take a more nuanced view, simultaneously defending the moral value of disabled people's lives, but also recognizing"—and I would add, debating—"the differential impact of impairment."[417:671]

Instead of seeking a "one size fits all" judgment on whether disabilities are socially constructed or whether their source points ultimately to some inherently limiting medical condition, what could be useful—though admittedly this also risks being politically divisive—is a consideration of the particular *cases of disabilities* in their plurality of expressions and plurality of sources of functional limitation. In the case of Down's syndrome under discussion here, it would seem that the bulk of the weight of the relevant evidence indicates that what is disabling about the impairments associated with this condition is by and large socially constructed and much (though not all) of it is socially remediable. There is no doubt that persons with Down's syndrome, as part of the broader populace of disabled persons in the UK, are critically disadvantaged, but this too is not the whole story. As Hull criticizes: "Conventional views of disability, while acknowledging the social manifestation of many disadvantages, tend to assume that since they would not arise in the absence of functional limitation, then those disadvantages must, and unproblematically, originate in functional limitation."[397:203] But, so long as persons with Down's syndrome (and their families) are included in the major gap between the disabled and nondisabled in terms of education,[418] income,[419][lxxiii] housing,

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[lxxii] Elsewhere, Shakespeare poses the question: "For example, if Down syndrome is just a social construct, then does that imply that the various medical and surgical interventions to help people with Down syndrome should be dispensed with? My friends with Down syndrome have variously suffered major cardiac malformations, gastro-intestinal problems and hearing loss. I understand that they have a higher likelihood of experiencing Alzheimer disease in later life. All these dimensions of the syndrome, together with the intellectual impairments which they also experience, result from having three copies of chromosome 21. None of this means that David, Charlie, Amy, Billy and the rest are less worthy of respect or inclusion. I count myself privileged to have known them. But I wonder how much it helps them to pretend that they are simply different, rather than disabled."[416:13]

[lxxiii] Research from the United States has shown significant correlations between reduced income for families of all types with a member with an intellectual or developmental disability as compared with controls from the general population.[420;421] The differential was even greater for single-parent households.[420-422] What is more, "[t]he proportion of single-parent households among those with intellectual disability–developmental disabilities is twice that of U.S. family households, with the great

employment,[400;see 423] social participation,[234;384;388][lxxiv] access to healthcare services,[243;424;425] and even respect for one's human rights,[426] then it makes no sense to assume that Down's syndrome as a genetic condition is mostly responsible for the resulting suffering, diminished quality of life, and "depressed" life prospects of these people.

Further examples of structural disadvantages that "weigh down" this population are the challenges faced through unemployment (or underemployment). For instance, a report entitled, "The Invisible Workforce" by the UK Down's syndrome Association, along with other anecdotal evidence, shows that prejudice[324] and social discrimination (often driven by fear, ignorance and misunderstanding) as well as inadequate access to public transportation stand out as common barriers to the workplace as experienced by persons with Down's syndrome.[see also 324;427] Of the approximately 15,000 persons with Down's syndrome of working age (18-60 years) in the UK, only 18% are currently in paid employment.[428] Such is the case despite the fact that in response to a survey posted by the UK Down's syndrome Association, two-thirds of those polled who were not working expressed that they would prefer to be employed.[429] According to Sue Buckley, a leading UK Down's syndrome expert: "With appropriate education, training and support some 60-70% of adults with Down's syndrome could find some form of employment."[lxxv;322] Given that non-participation or marginalisation within the labour-market is a primary index that tracks social stratification in most industrialized societies, the resultant compromises to quality of life and life prospects, even to the point of threats to social identity, are serious. As Richard Jenkins observes: "In a society where active citizenship for those other

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majority being headed by women; the economic disadvantage in these households was found to be large…"[419:223]

[lxxiv] In a qualitative research project, Alderson notes that adult interviewees with screenable genetic conditions (including five individuals with Down's syndrome) recounted experiences of "frustrations, pains and restrictions of prejudice. The …interviewees tended to attribute problems to negative attitudes and social barriers rather than to their congenital condition, and most were frustrated at not having the opportunities, employment, income and social acceptance to enable them to live their lives as fully as they thought they could." Alderson further observes that: "Research which enquires beyond morbidity into people's potential and achievements, and the social influences which support or constrain them, is required before their ability can be assessed realistically. …Until far more research is conducted which takes proper account of social contexts, generalisations cannot be formed about the costs of unavoidable dependence, or the value and quality which people with Down's syndrome experience in the everyday details of their lives."[324:635-636] Cf. Taylor[395] for further explanation of relevant, chronic disadvantages in disabilities populations within the United States.

[lxxv] Admittedly there is a dearth of current research on this point. In 1994, Ward and colleagues noted an unresolved paradox "in which, implicitly and explicitly, expectations of employment are raised in the minds of those with such needs, whilst at the same time they are engaged in a continuing round of training, followed by more training, with actual employment being postponed as a consequence of a combination of factors, including, for example, limited opportunities, the general economic recession, and possible employer resistance" (quotation from Thomson *et al.*[430:329] citing Ward *et al.*[431]).

than the very rich is associated with individualistic notions of 'earning your keep', the perceived inability to do so poses a problem in terms of one's overall social membership."[432:557][lxxvi]

This is not to deny that there are cases where individuals with Down's syndrome sustain impairments by way of their genetic endowments that have significant, determining, negative consequences for their lives and the lives of their families. The degree of impairment cannot be predicted on the basis of PGD testing in advance of birth: as such, the risk of "serious" impairment does exist. But as Jackie Leach Scully points out:

> Since no society ...has entirely eliminated the social or cultural contributions to disablement, the magnitude of the intrinsic disadvantage is only really clear when it is so overwhelming that it renders the other contributions trivial.[lxxvii] For many phenotypic variations, it is not yet possible to assess how much intrinsic disadvantage would be left over if social responses were different.[74:172-173]

Thus, only with a better grasp of what the experience of living with Down's syndrome is like (both from the perspective of individuals with Down's syndrome themselves as well as their family members) and what it *can* be like under changed conditions—only then will we be on firmer grounds on which to base judgments about intrinsic and modifiable disadvantages associated with Down's syndrome.

Undeniably, a life with Down's syndrome—as also for a life with a family member who has Down's syndrome—is one that forecloses certain opportunities. For persons with Down's syndrome, these closed opportunities are in part on account of limiting factors of their particular genetic endowments. Here, however, what needs to be stressed is not that this fact proves an inherent disadvantage as attaching to the syndrome. The question is whether these

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[lxxvi] Jenkins explains that: "The important point to stress here is not that most people with mental handicaps cannot work in open employment – regardless for the moment of the kind of work involved, we know that they can[433-436] – but that, due to a variety of constraints, they do not. These constraints include: restricted employment goals on their part (and their families), employer and employee discrimination, a shortage of the required specialised support resources and, in the UK, a community care policy which makes many people with mental handicaps dependent on state-funded private residential care which they could not afford if they were in manual or other low status employment. ¶People with mental handicaps, if they are economically active, are marginal workers in the labour market. For most, however, their marginality is more general: their total exclusion or absence from the labour market is an expression of this global social marginality, rather than *vice versa*. The crux of the problem is the nature of their membership of society, the degree to which they are citizens, and, in the final resort, their adequacy as human beings in the eyes of their fellows. This raises the question of whether they are 'in' the stratification system at all."[432:571]

[lxxvii] See also Pogge[437] and Silvers.[438]

limits have to, or are even likely to, impact negatively on those lives, and if so, for what reasons? In this regard, several points are relevant. First, the fact of limitation of opportunity in any human life is hardly abnormal. It is true of any life course.[400:38-52;439] Second, rather than assuming this truncation of opportunities to confer a necessary loss on the part of persons with Down's syndrome, Asch and Parens (more generally speaking about disabilities) as well as Wong[310] (speaking specifically about Down's syndrome) would have us consider instead the near infinite range of remaining opportunities available through seeking alternative modes of the same or similar activities.[171:43]

There is also the feminist intersectional concern, "For whom is a given condition considered 'serious' and in what context this is the case?" In this regard, there is reason to believe by way of the evidence given above, that environmental conditions as well as social prejudice have made for structural disadvantages constricting the development and curtailing life opportunities for persons with Down's syndrome as well as their families. Prejudicial stereotypes and general negative social attitudes[lxxviii] against persons with Down's syndrome are now strongly entrenched within prevailing social norms not only in the UK, but throughout (at least) the Western world.[227;443] And as feminist bioethicist Susan Sherwin has wisely pointed out, "the greatest danger of oppression lies where bias is so pervasive as to be invisible."[98:10] So also, another prominent feminist bioethicist Marsha Saxton, points to the realities of social stigma[444;445] when noting that: "One's options in life as a person with a disability may indeed, in some ways, be limited, but oppressive social conditions have so distorted the public's

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[lxxviii] Saxton[174] gives examples of some of these negative attitudes and how they are experienced more generally by members of disability communities. Nelson[339] explains how disability activists take issue with medical systems that exaggerate the burdens of disabilities and underestimate the functional abilities of disabled persons. This is compounded in medical language that "reinforces the negativity associated with disability by using such terms as 'deformity' or 'defective embryo or fetus'."[339:73] Further examples of negative language are to be found on the HFEA's website explaining preimplantation genetic screening, under the tab "Treatment and Storage Options" for prospective clients. There, the question is asked, "Is PGS for me?" The first answer states: "Your clinic may recommend genetic screening of embryos, particularly if you are over 35 and have a higher risk of having a baby with a chromosome *problem* (such as Down's syndrome) [emphasis added]."[440] Moreover, a research project approved and listed on the Authority's website tells of a new technique preimplantation genetic haplotyping (PGH) which reportedly "improves the reliability of single cell genetic testing and enables it to be extended to a wide range of diseases." Work is underway to see if this approach can be extended to include tests for what is described as "some *serious* or lethal chromosomal trisomies such as Edwards, Patau and Down syndrome [emphasis added]."[441] Though a conceptus affected with trisomy 18 (Edwards syndrome) or trisomy 13 (Patau syndrome) rarely result in live births and those that do result have low rates of survival even past a few days, this is not true for trisomy 21 (Down's syndrome). Even with respect to trisomy 13 and 18 a recent study by Janvier and colleagues (response rate 67-87%) reported that "despite their severe disabilities, 97% of parents [to 272 'affected' children] described their child as a happy child. Parents reported these children enriched their family and their couple irrespective of the length of their lives."[336:293] See article by GDSF, "Words Can Hurt".[442]

perceptions, as well as how disabled individuals might internalize these perceptions, that it is difficult to assess the true impact of disability on the individual's life experience."[lxxix;174:150]

Thus, when considering the appropriateness of PGD for Down's syndrome it is important to consider what else besides genotype may be playing a significant role in fostering the *social perception* that this is a "serious condition." What is more, to more fully appreciate some of the sources of disadvantage associated with Down's syndrome it is necessary to expose the ways in which society has constructed such a narrow conception of which opportunities are recognized as valuable for a "life with quality" and to resist the sometimes irresponsible reductionist narratives about the finalities of genetic determinations.[lxxx309] Parens and Asch point out that unfair "discrimination results when people in one group fail to imagine that people in some 'other' group lead lives as rich and complex as their own."[171:41] It is important to note, as Martha Minow [108;447] points out, that many differences between individuals or groups are significant if a certain sector of the population in a culture succeeds at vindicating their significance in the eyes of others. This in turn creates hierarchies of value that track what are taken to be more and less desirable traits in a given culture or society.[108;447] As feminist disability theorist Susan Wendell explains:

> If you are 'other' to me, I see you primarily as symbolic of something else—usually, but not always, something I reject and fear and that I project onto you. We can all do this to each other, but very often the process is not symmetrical, because one group of people may have more power to call itself the paradigm of humanity and to make the world suit its own needs and validate its experiences.[lxxxi] Disabled people are 'other' to able-bodied people, and ...the consequences are socially, economically and psychologically oppressive to the disabled and psychologically oppressive to the able-bodied.[414:271]

[lxxix] This puissant criticism from Saxton is directly relevant for research on quality of life assessments and for investigations into the association of disability and suffering mentioned above.

[lxxx] Epidemiologist Abby Lippman notes: "Social conditions are as enabling or disabling as biological conditions. Why are biological variations that create differences between individuals seen as preventable while social conditions that create similar distinctions are likely to be seen as intractable givens?"[446:362]

[lxxxi] Wendell includes the following note: "When Simone de Beauvoir[448] uses this term to elucidate men's view of women (and women's view of ourselves), she emphasizes that Man is considered essential, Woman inessential; Man is the Subject, Woman the Other. Susan Griffin[449] expands upon this idea by showing how we project rejected aspects of ourselves onto groups of people who are designated the Other."[414:276,nt.12]

Explaining how oppressive forces entail negative consequences to both parties of vicious, and commonly spurious, dichotomies[lxxxii] (e.g. able/disabled, male/female, etc.) need not detain us here. The important point to note is that for persons with Down's syndrome to carry the label "disabled", amongst other things, prevalently is taken to mean that they fall short of the ideals recognized by the majority, and thus cannot measure up to the norm of what Daniels has denoted as "species typical functioning".[451] In the face of widespread prejudice—sometimes subtle, and residual—as well as entrenched stigmas against persons with disabilities, as feminist bioethicists Asch and Gail Geller point out: "...people who perform physically, cognitively, or psychologically below the ideal and the average are likely to experience limitations and hardships even after current social, legal, and financial obstacles are removed."[lxxxiii;168:330]

In this regard, Crossley and Shepherd (along with certain feminists[414;452;453]) have pointed to an overvaluation—or, very narrow conception—of the principle of autonomy, so esteemed in modern Western societies[454] that has led to modelling "ableism" as independence from others' care.[455] This exclusionary interpretation of what autonomy could mean for living a human life, contributes to making for social attitudes that prove inhospitable to disability traits and conspicuous relations of dependency.[456:xv][lxxxiv] Persons with Down's syndrome and their supportive parents who may rely on (or, perhaps even prize) relations that involve dependency, or interdependency,[459;see 460][lxxxv] face discrimination on this account in educational systems and work environments that presume more conventional ideals of autonomy

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[lxxxii] Drawing hard and fast distinctions between able-bodied and disabled, cognitively capable and cognitively impaired persons, though often appealed to in making sense of difference—*e.g.* when these distinctions may be important to determine who is legally entitled to certain benefits on the basis of these differences—can also work in ways to hide from view the fact that most impairments are scalar. Shakespeare gives the example that there is no polar distinction between being blind and being sighted, but rather gradations of visual acuity. Welfare systems may define how much visual impairment qualifies someone to be declared legally blind as necessary for official and administrative purposes. Shakespeare states: "Therefore there are processes of social definition and meaning inherent in the categorisation of impairment, and in the identification of disabled people. Equally, the ways in which societies define and categorise physical and mental differences have implications for the way in which people with those differences live their lives."[416:13;see 450] Minow adds that: "[R]elational insights show a mutual dependence between 'normal' and 'abnormal' people, and between male norms and women who do not fit them. Hierarchical pairings that deny the mutual dependence of both elements provide clues to deeper motives that make oppression and degradation seem natural and inevitable. Such patterns deposit on some people the fears of all of us about vulnerability and danger."[108:219]

[lxxxiii] In the original, Asch and Fine reference the following: Asch.[167]

[lxxxiv] See also Alderson,[324:628] Davis,[457] and Wendell.[414:271;458]

[lxxxv] See also Kasari and Freeman.[459] On a related point, Wendell notes that: "some disabled people spend tremendous energy being independent in ways that might be considered trivial in a culture less insistent on self-reliance; if our culture valued *interdependence* more highly, they could use that energy for more satisfying activities [emphasis added]."[414:273]

(understood as non-relational).[400] Sophia Wong (herself a sibling to a brother with Down's syndrome) echoes this challenge, saying: "If we can move toward overcoming the Enlightenment fetishization of cognitive ability and dislodge the institutional barriers enforcing cognitive difference, perhaps we can build a society more welcoming of persons with Down's syndrome."[310:115]

As the evidence from this chapter thus far shows, there is no good reason to assume that life with Down's syndrome *has to be* or is even *likely to be* a life of diminished opportunities, suffering, and low quality for either persons with Down's syndrome or their families,[461] except (for the most part) insofar as environmental and social conditions make it so.[lxxxvi] Having a child with Down's syndrome is not necessarily regrettable—in fact, it's not even likely to be regrettable.[355;364][lxxxvii]

Rehabilitation psychologist Beatrice Wright aptly points out a common misconception by which it is assumed that an impairment resulting in some incapacity in one area of life will spread to incapacity *writ* large for the rest of one's life, including the lives of one's close relations.[462] Asch points out another prejudice against life with disabilities in the common assumption that there is no benefit to offset the "burden"[lxxxviii] of extra care that comes with raising a child with some kind of impairment.[463:1653] As the above evidence shows, families of persons with Down's syndrome find definite benefit and value from having a family member with Down's syndrome. The need to accept trade-offs and to realign personal or familial expectations when raising children is a very routine experience that most parents face when taking into account the abilities of their children. Suffering in the form of compromises to well-being are to be expected, while at the same time not to be underestimated in raising a child with Down's syndrome.

As we have seen, the likelihood and extent of disabilities associated with Down's syndrome are a result of the interplay of individual impairment and contextual factors.[75] Assumptions of a simplistic cause and effect relationship between Down's syndrome and individual and family suffering[402:92;see 407] are not borne out in a review of the literature.[see 464] The experience of

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[lxxxvi] See, furthermore, Nelson[339:72-73] as well as Saxton.[174]

[lxxxvii] For example, Nelson reports that disability rights activists have pointed out that not all individuals with Down's syndrome or their parents live painful, frustrated, or tragically diminished lives. The same is true for individuals with spina bifida, cystic fibrosis, Fragile X, and other genetic disorders.[339:67]

[lxxxviii] It is conventional to speak of the care of persons with disabilities as being a "burden" (e.g. talk of "burden of care"). This negative presumption might better be avoided by referring instead to the additional "responsibilities" of care, rather than presuming that these responsibilities carry a negative value.

families with members who have Down's syndrome is variable,[lxxxix] and a "pathological response" is not inevitable.[465] The argument here is that the experience of social discrimination as well as neglect, prejudice, stigmatisation, segregation, and a general history of disadvantage have done much to constrict the life opportunities of persons with Down's syndrome. These prevailing negative conditions and social circumstances have been, and continue to be, dominant factors in their own and their families' experiences of (and expectations for) suffering, notwithstanding much evidence of resilience.[400:34]

Summary of chapter conclusions

In summation: in this chapter I have argued that according to the factors for assessment given in the Code of Practice section 10.7, approving applications for the use of PGD for Down's syndrome is (considering the evidence) problematic. Those aspects of the syndrome which result in compromises to health can in most instances be successfully managed (or treated) through the provision of adequate medical care, while other aspects can be addressed through the provision of adequate social supports and provisions for living. The intellectual impairments associated with Down's syndrome have been exaggerated and the evidence points to systemic gains in terms of the intellectual and social capacities of persons with Down's syndrome by way of educational and other kinds of social reforms or interventions. The evidence brought forward here also argues against any presumption that either persons with Down's syndrome or their families are likely to have lives dominated by suffering. In fact, most suffering and disadvantage associated with Down's syndrome for the affected individuals and their relatives is either the product of social conditions (including, most notably, prejudice, stigma, and social discrimination) or can be significantly alleviated or ameliorated by way of changes to family circumstances, the provision of social supports, and various forms of societal reform.

In making sense of Down's syndrome according to the theoretical models of disability, it seems that not all or even most of the impairments associated with Down's syndrome necessarily result in disabilities (*à la* the social model of disability); many, instead, result in disadvantages. I have argued that it still makes sense to uphold a distinction between disability and disadvantage (*pace* strong versions of the social model).[397;399] By doing so, I am leaving

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[lxxxix] Tom Shakespeare has drawn attention to the fact that "the differences between disabled people are as important as the similarities: for example, examination of the role of class is paradoxically absent, even from materialist disability studies."[75:198] This again is indicative of a need for more on point research in this area. See Jenkins.[432]

room for the reality that even under the most supportive and tolerant social conditions, some impairments associated with Down's syndrome would still result in disabilities (*à la* the medical model). This is to say, the disabilities resulting from such impairments would not be exactly like the sorts of disadvantages resulting from, say, gender which would arguably be completely remediable by way of altering social conditions.[cf. 74;397] Even so (to repeat), as long as persons with Down’s syndrome (and their families) are included in the major gap between the disabled and nondisabled in terms of education, income, housing, employment, social participation,[234;384;388;see 395] and access to healthcare services—and as long as they are the object of widespread oppressive forces, then it does not make sense to assume that Down’s syndrome as a genetic condition is itself inherently (or, "in principle") disabling and mostly responsible for the resulting suffering, diminished quality of life, and compromised life prospects of these people (*pace* strong versions of the medical model). Most of the challenges associated with Down's syndrome as a disability are in fact contingent on situational factors, against which the resulting disadvantage could be alleviated significantly, if not done away with altogether, in environments with different social structures, supports, and values.[309]

Finally, I have been concerned to point out that judging Down's syndrome to be a "serious" genetic condition because of its negative potential to cause suffering and to compromise quality of life for individuals with this condition or their families is very problematic. Justification(s) for extending PGD testing for Down's syndrome under the current regulatory instruments as an appropriate application of this technology seems not to follow from a consideration of the evidence as given in the relevant literature concerning the capacities and quality of life possible for most persons living with Down's syndrome. Thus, under the current UK laws (the HFE Act) and the HFEA's guidelines (Code of Practice), licensing PGD testing for Down's syndrome as an appropriate use of this technology would seem to be very problematic and, as we shall see in the next chapter, very misleading.

Chapter 3: Considering some structural problems with UK regulation of PGD

Chapter objectives and outline

The previous chapter pointed to the fact that: i. there is suffering and disadvantage associated with Down's syndrome, both from the perspective of those directly affected by life with this condition as well as from the perspective of their family relations, but ii. that much of the experience of the disability is rooted in environmental and social sources, and much of this is remediable through the provision of modern medical, educational, and (more broadly) social supports and services. As stated in chapter 1, a power-focussed *feminist* approach to bioethics demands that we take seriously the role that health law and public policy assume in relation to general structures of oppression in society. This chapter follows up on some of the insights of power-focussed feminist bioethics insofar as it will attempt to expose some of the structural and institutional forces of oppression that lie in the way and impede the necessary social-cum-moral progress required to adequately address not all, but some important ability needs of this population.

As bioethicist Susan Sherwin points out, feminist anti-oppression theorists are especially concerned to understand inequalities deriving from problematic relations and dynamics between social groups, not simply between individual members within given groups.[90;106:83] In making the investigation of group-based power differentials central to the agenda of doing feminist bioethics, Sherwin proposes "that we always interrogate [health] practices or policies to determine how each contributes to existing patterns of oppression", especially those that affect disadvantaged social groups. Here we need to query whether the established health practices and polices worsen or exacerbate current inequalities, improve the situation, or remain neutral with respect to social group differences. As Sherwin concludes: "we [also] need to ask whether there are alternative practices or policies we might adopt that would make a larger contribution to reducing social inequalities."[106:84]

Following Sherwin's exhortation, this chapter considers some of the ways in which the UK regulatory system for PGD threatens to compound the risks of prejudice and institutionalize social discrimination against persons living with Down's syndrome—all forces of disempowerment of this social group. First I consider why the relatively recent move of the HFEA to have an exclusive list of "serious" genetic conditions[64] for which PGD testing is approved "in principle" contributes to negative forces of oppression against this social group. In sum, my challenge to the principlist approach to regulation according to (supposed determinations of) "seriousness" is that it tends to place the emphasis on the condition itself[xc] rather than looking to situational or environmental

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[xc] Speaking to this issue, in a qualitative study which interviewed AHR professionals, Counsellor 28 observed: "'I've always thought, perhaps it's one of the reasons it hasn't been done in law, is that you can't kind of ... write a list of things or conditions that you consider serious ... And I think one of the reasons it's not been done in law is that it - the perception and the reality of seriousness isn't just about

factors implicated in disability. Furthermore, this approach to regulation in effect overlooks the complexity of the involved contingencies of associated disability for a life with Down's syndrome. The final section of this chapter attempts to build back from the particulars of the study example (case) of Down's syndrome to consider some of the broader disability concerns at stake in limiting access to PGD based upon the HFEA's list of what are determined to be "serious" genetic conditions. I end the chapter by suggesting reasons for dealing with this situation by expunging the list altogether.

Problems with the HFEA's new list of "in principle" serious conditions

To understand the gravity of the risks of harm against persons with Down's syndrome that attend to having Trisomy 21 listed as one of an exclusive set of conditions considered to be "in principle" serious according to the HFEA, we will need to look at the evolution of certain recent revisions to the HFEA's licensing procedures in response to the changes to the HFEA Act 1990 (as amended, 2008) mentioned at the beginning of chapter 2.

In November 2001 the results of a public consultation on PGD were released by the HFEA and Human Genetics Commission (HGC). At that time, the overwhelming majority (80%) of the public polled agreed that "the seriousness of a condition should be a matter of clinical judgment based on general guidance."[29] Alternatively, 20% of those polled felt that the seriousness of a genetic condition should be determined by referring to a list of specified conditions. Reportedly, the latter responses "tended to see a danger in allowing *individual preference* to be a determining factor in the use of PGD [emphasis added]."[29:Part 2, p.15, para.20] Also at the time, the Joint Working Party (of the HFEA and HGC) "agreed that the guidance given on the use of PGD should not comprise a prescriptive list of 'serious conditions' for which the use of the technique was thought to be appropriate."[29:Part1, para.23] They recommended that guidance should "support difficult parental choices rather than appearing to discriminate against individuals with certain conditions."[29:Pt.1, para.22][xci] Since then, rather than regulating access to PGD according to an "objective list" of licensable conditions, HFEA guidance has been oriented towards emphasizing the importance of the views and experience of those seeking PGD for determining what counts as a significant risk of a serious genetic condition.[see 132:110;156:s.4.4]

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the condition itself.'" Rosamund Scott (from whom the quotation just given was taken) notes: "Having recounted the above case, Counsellor 28 continued: '[I]f you did have a list, I think you would be less of a clinician, because the notion of clinical judgement and care of your patient or family would have to go out the window.'"[176:214]

[xci] See also the HFEA & HGC (UK).[29:Part 1,para.23]

However it is now a statutory requirement[466:s 1.,1.1-1.4] under the Act (as amended 2008) "that the HFEA [itself] must be 'satisfied' that PGD is carried out only where either there is a significant risk that a child to be born will have or develop a serious condition or where a physical or mental illness or other medical condition is gender related" and for which it can be screened against on this basis.[467:s.1.3] Counsel to the HFEA has determined that:

> whilst the statute does not require the Authority to satisfy itself as to the risk and seriousness of genetic conditions in every *individual* case, it does require the Authority to determine significant risk and seriousness, in general, for each genetic condition for which PGD is offered. There is no need for the Authority to base this decision on the facts of an individual case, even for the first use of PGD for that condition [emphasis in original].[156:s.5.3]

On the basis of this Counsel, the HFEA's Head of Research Regulation (O'Toole) and Head of Policy (Tizzard) put forth in an *Authority Paper* (18 March 2009) that: "The Authority could, therefore, agree in principle that PGD could be used for a particular genetic condition, based upon 'the nature of the disability, illness or medical condition which is likely to result from that abnormality *in general* [emphasis in original].'"[156:s.5.3]

As of October 2009, the HFEA has proceeded to implement a two-tier level of assessment for determining appropriate grounds for accessing PGD treatment. Rather than the attending ART clinician (in consultation with their client) needing to be satisfied of the significance of risk and seriousness of a condition,[155:5] it is the Authority itself that "needs to ascertain that the statutory requirements for risk and seriousness are principally met for a given condition." It is understood that the clinician or centre is then "tasked with ensuring that PGD is only offered in cases of significant risk of a serious condition."[468:s.3.19] The HFEA Licence Committees have thus published a list of select conditions (which includes Down's syndrome) that can, in principle, be considered "sufficiently serious" to warrant PGD testing.[64]

In the past, conditions were approved on the basis of requests from clinics on behalf of concerned clients. The HFEA has made a point that the current list of approved conditions for PGD is not necessarily client driven: i.e. the list is not to be misconstrued as representative of already approved applications. More specifically, treatment centres no longer apply for variations to their licences to carry out PGD for particular couples. Now instead, licensed centres may carry out PGD for any of the conditions listed on the HFEA website.[64][xcii] Unlike in

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[xcii] The list, according to the HFEA, serves as "an informative reference point for clinics and patients." Obtaining IVF and PGD services can be a notoriously slow, involving process given the many barriers to

the past, when applying for a condition not previously licensed, centres are no longer required to apply to the HFEA on the basis of a particular patient case. Instead, they can make an in principle application for a given condition, and the HFEA makes an in principle judgment about that condition.[469]

Given the evidence of the previous chapter that so much of what makes Down's syndrome disabling is contingent on environmental or situational factors, including elements that are outrightly socially-induced[xciii] (for example, all the problems associated with stigma[444;445]), it seems somewhat of a strain to consider that there is some in principle "nature of the disability, illness or medical condition which is likely to result from that abnormality *in general*"[156:s.5.3] that makes it a "serious condition."[xciv] As Hull points out, to underestimate the socially-induced

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access: for example, financial costs and associated travel time due to the limited number of clinics in the UK. Publication of licensable PGD conditions, besides meeting the goals of transparency, may also speed up the process of obtaining PGD services for any given couple.[469]

[xciii] This thesis has shown that much of what makes for the hardships associated with Down's syndrome is context-dependent and value-laden. Certain critics may protest here, claiming that "socially constructed" disabilities can be "serious." However, *knowing* what is meant by saying that Down's syndrome is "serious" is unclear. As given in UK law and regulation the term "serious" remains vague. If what is being asked here is whether socially-induced disability results in real harm or losses of functioning that adversely affect capacities for flourishing, then much of the evidence given and argument put forth in this thesis would support this claim. Indeed, the fact that real harm is involved with socially-induced/socially-constructed disabilities and that these harms are rooted in unfair treatment of this population makes the redress of this situation a matter of social justice as indicated in chapter 1. But owning the harm as real, even if it is socially constructed and socially remediable, is not equivalent to labelling such disabilities as "serious" according to the indeterminate meaning this standard carries in the context of UK PGD regulation. As we have seen, the relevant law and policies do not suggest a clear meaning for serious and in this way fail to account for the possibility that disadvantage, which some may view as "serious", is socially constructed yet remediable in various ways. In my view there needs to be a public conversation *if* seriousness is to be the standard. This standard might be relative but there would be value in knowing how it means what it does. (I am indebted for this last point to Lynette Reid).

Though it goes beyond the scope for this thesis as an internal critique, another consideration is whether it is reasonable and/or ethical to use PGD to avoid these social harms for one's prospective child or for oneself as a parent—especially, if those social harms are rooted in oppression. In this regard, though, UK law is clear that using PGD for social sex selection, for example, is absolutely prohibited.[144:Sch.2,s1ZB] What is more, the HFEA is now in the process of considering de-listing conditions which have since the time of their original approval become treatable.[470;471] It would seem then that if the harms attached to a condition are rooted in social conditions and can be "treated" by way of medical and social interventions as with Down's syndrome, this argues against the legitimacy of using PGD in such a case on the basis of UK law and policy. I say this, however, knowing that there will be disagreement across interpretations over what counts as a treatment with sufficient therapeutic remediation and one which it is reasonable to expect will succeed in achieving this outcome by way of the proposed therapeutic intervention.

[xciv] In an Ethics and Law Advisory Committee discussion paper from 20 January 2009, Chris O'Toole (HFEA Head of Research Regulation) stated that: "Licence Committees use the criteria which clinicians should use when deciding whether to apply for a variation to their licence to carry out preimplantation genetic testing for a particular couple at risk of passing on a particular condition."[472:s.2.1] These are the criteria given at G.12.3.3 in the Code of Practice (7th edition)[473] which are virtually the same as those

factors of disability, "in the language of Lukàcs,[xcv] is to suffer from a reified view of disability whereby disability is mistakenly viewed as being natural and fixed."[397:208]

The HFEA does acknowledge that *clinicians' judgments*—notice not parents' judgments—are needed to assess the particular suitability of PGD in each case, but the shift in authority and emphasis is nonetheless significant. It is to be remembered that no applications of PGD are permissible in the UK other than those which would screen for conditions given on this list. To be on the list, the conditions are understood as specifically warranting "treatment services" in the form of a medical, surgical, or obstetric intervention.[144:s2(1)] In this regard, Wasserman and Asch have argued that the practice of singling out and designating a special class of genetic conditions as "serious" and therefore appropriate to test for, holds the risk of giving medical imprimatur to some of society's strongest fears and prejudices about what it is to relate to, live with, and support persons with these conditions or other disabilities.[477] As Asch asks: "Why should parents be told by test designers: 'We think that cystic fibrosis, or muscular dystrophy, or deafness, or Down's syndrome should make parents think at least twice before contemplating childraising; but other conditions are too trivial for parents to object.'"[478] According to Asch and Wasserman, "a policy that promotes selection against embryos with disabling traits conveys the strong impression that the problem is the disability itself rather than the society that could do so much more to welcome and include all its members."[xcvi;477]

This criticism may seem too strong: after all, the HFEA would claim that it is legally permitting and licensing, not promoting, selection against embryos affected by "serious"

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referenced in s 10.7 of the Code of Practice (8th edition).[121] In an Authority paper from 18 March 2009 Chris O'Toole (HFEA Head of Research Regulation) and Juliet Tizzard (HFEA Head of Policy) stated: "Once the application is made, the Authority uses the same criteria to decide whether or not to approve the centre to offer PGD for that genetic condition. If permission is given, then the centre may offer PGD for that condition to any other patients it sees fit, subject to the criteria in the Code."[156:s.3.4] It is against these criteria then that it would appear difficult to see how the judgment is valid that Down's syndrome is an appropriate application for PGD on the standards given in the HFEA's Code of Practice. Presumably, those defending the HFEA could draw attention to the fact that the new licensing procedure is working to develop new instruments[467;468] with which to determine the "in principle" appropriateness of using PGD. According to a statement by Danny Edwards (then HFEA Policy Manager): "Guidance exists for the Licensing Committee regarding establishing seriousness."[468] It would be not only useful, but also important, for the purposes of maintaining its responsibilities for transparency, openness, and a commitment to objective decision-making,[474] if the HFEA published these criteria—which presumably are supposed to be indicative about what is objectively "serious" about any condition for which PGD testing is considered "in principle" appropriate—in the Code of Practice. Doing so would also help to meet the "intent behind the updated legislation to ensure that the licensing process for PGD is set out within clear boundaries."[475:5]

[xcv] See Lukács.[476:83]

[xcvi] With respect to PND for disabling conditions, Mary Johnson makes a related point saying: "A disabled foetus represents for parents a problem that may have far more to do with society than with disability. Disabled children [oftentimes] confront a hostile environment."[479:14]

conditions. When thinking of the UK regulatory regime for PGD, designed as it were according to the recommendations of the Warnock committee to make ARTs legally controlled activities, we often think of what law and policies do to limit or stop certain uses of these technologies. Less often do we think of what choices are (perhaps unwittingly) being promoted by way of legal authority. As said before, it is important to bear in mind the legal context in which PGD is treated as an otherwise banned activity but for those conditions judged as "serious" or "serious-enough" to warrant licensing. In this (over-determining) context, by demarcating this special class of genetic conditions and labelling them "serious" thereby judging them to be uniquely appropriate and therefore licensable for PGD testing, this line-drawing approach to regulation risks making it seem that selecting against having a child with a "serious condition" (e.g. Down's syndrome) is *prima facie* more legitimate—as validated by the authority of medical science and invested, ultimately, with the foundational authority for such judgments provided by the State with the HFE Act 1990 (as amended 2008)—than choosing to resist this established, widespread tendency in society.[477] These laws and policies are therefore at risk for compounding public stigma against persons with Down's syndrome and their families. Such promotion of systematic bias needs to be named for what it is: namely, oppression[106:83-84]

Defenders of the HFEA's approach here might argue that since the degree of impairment cannot be predicted in advance of a prospective life with a given genetic condition, an "in principle" decision to allow PGD for say, Down's syndrome, would be justified as taking account of the risk of serious impairment in, albeit, a small number of cases that are no less "severe" for being rare. And, members of the HFEA 2006 Ethics and Law Committee are said to have "recognized that if a condition is serious then any risk is significant".[480] There is certain merit to this reasoning, and my position is such that I would definitely not wish to challenge prospective clients who are "risk averse," so to speak. To repeat: my issue with this principlist approach to regulation according to the criterion of "seriousness" (as given) is that it tends to place the emphasis on the condition itself rather than looking to environmental factors, thus overlooking the complexity of the involved contingencies of associated disability for a life with an extra chromosome 21. In this regard, the continuous focus on the genetic quotient[481] to factor the effects of disability propels us to bypass the important question of whether the impairments that come with "serious conditions", in this case Down's syndrome, *have to be* or are even *likely* *to be* disabling, and to what degree social conditions[xcvii] are the primary factors resulting in associated disabilities.[xcviii] The burden of this thesis has been to force a pause in answering this question to consider the evidence. Weighing the evidence, I want to suggest that most (though not all) of what is debilitating and limiting about living with Down's syndrome (or being in a family with a member with Down's syndrome), has been in the past, and importantly still is, either exacerbated by, or the direct product of prejudice, stigma, and unjust discrimination and can be redressed by way of social, political, and legal reforms.[xcix;477;482;483]

The argument could be made that even though many negative aspects associated with living with Down's syndrome (as an individual) or caring for a Down's syndrome child (as a parent) can be addressed through the provision of adequate social supports, historically speaking, there is a long standing practice of inadequate social and material provisions to persons with Down's syndrome as well as their families. Given this context, what is the HFEA to do in the meantime? Knowing that there "will"[c] be inadequate supports and services for people living with Down's syndrome (and their families), what weight should be attached to the fact that the effects of a disability could be ameliorated by adequate support when making a judgment about the "seriousness" of that disability?[ci] In responding to this question, it is important to re-emphasize that the language of "seriousness" as promulgated by the amended HFE Act (2008) and used in HFEA policies in these matters, tends to place emphasis on the "in principle"—understood as, in general, inherently incapacitating—nature of the impairments that come with a given condition. The criticism just stated, then, actually underscores the situational and social rather than genetic character of the disabilities which persons with Down's syndrome and their families face.[cii]

[xcvii] Wendell notes that "[m]any of the 'special' resources the disabled need merely compensate for bad social planning that is based on the illusion that everyone is young, strong, healthy (and, often, male)."[414:265]

[xcviii] As Asch notes, most impairments become disabilities because of the prevalent "mismatch between the range of people actually in the world and the institutional practices, physical structures, modes of communication, and social attitudes that assume a much narrower range of human beings than exist."[478:318]

[xcix] See Waddington[400:33-38] and Scott.[325]

[c] These sorts of predictions are not without problems as they imply that the lack of supports and services is inevitable.

[ci] I thank an anonymous reviewer from the journal *Medical Law Review* for this criticism.

[cii] The pragmatic concern of where adequate material, social, and political support is to be found to address the needs of persons living with Down's syndrome and their families, is of course valid and the position of this thesis is not to dispute the extension of PGD (*per se*, just the extension of PGD on the basis of UK law and policy) for those who wish to utilize this technology to prevent having offspring with Down's syndrome. Questions concerning the lines of responsibility for assuming the extra social supports and economic costs incurred in raising a child with Down's syndrome are also issues that need further

Critics of the position put forth in this thesis might further rejoin that the evidence given, which seems to normalize much of the experience of having and raising a child with Down's syndrome, reflects an exceptional ability of certain families to cope with the relevant difficulties, but that this is not evidence of the comparative neutrality of living with, or raising a family member with, Down's syndrome.[ciii] Retrospective judgments concerning whether the experience was rewarding (or worthwhile) as opposed to laden with suffering, are quite different from prospective judgments concerning what one might want to attempt to undertake. That many families with members with Down's syndrome are resilient and able to cope well (for the most part), does not prove it would be a mistake in reasoning[civ] (moral or otherwise[173;412;416:esp.13-14]) for prospective parents to want to avoid such challenges—challenges that may be perceived as holding too great a risk of suffering for the family, even if these challenges might be beneficial either on a personal scale or to family life more generally.[cv;171]

This criticism has certain force insofar as it makes sense of parents' preferences for what sorts of challenges they wish to take on, what kinds of children they want to have, and what their expectations are for the kind of family life they hope to have with their child or children. In response, I would say that the point here is not to argue against or in any way to judge or question the reasons prospective parents might have for wanting to use PGD services to select against embryos that carry a strong likelihood of yielding offspring with Down's syndrome. We do well to note, though, that in licensing PGD for Down's syndrome the HFEA does not understand itself to be making available a tool for clients to simply better guarantee the results

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exploration and research, but outside the scope of this thesis. Note that most literature that details information about the economic costs associated with Down's syndrome take an approach of cost-benefit analyses of proposed screening programmes.[417] See for example Gill *et al.*[484] and Hagard and Carter.[485] For a critique of the lack of economic and social support for prospective parents' choices to raise: i) children with disabilities, and ii) children with Down's syndrome, see respectively Shakespeare[417] and Alderson.[244]

[ciii] For a rejoinder to this criticism, see Asch[486:esp. 248ff.] and Parens and Asch.[173] Cf. Scott.[325]

[civ] Even though not all instances of Down's syndrome involve the sometimes feared problems associated with the genetic condition, the risk is real that any prospective child or adult with trisomy 21 may face a life coping with these feared challenges. A prospective parent who chooses to act on her/his conclusion that it is better for herself/himself not to have, or to be at significant risk to have, a child with this disorder is not necessarily acting unreasonably (and certainly not unethically) according to anything argued for in this paper. Cf. Krahn and Wong,[117] Steinbock,[412] Nelson,[339] and Scott.[325]

[cv] What is more, there is the possibility of selection bias in (some of) the studies where parents who have positive experiences to report concerning their experience of having a child with Down's syndrome are over-represented and those with negative attitudes and experiences remain unknown. This criticism should be checked with a meta-analysis review. And, insofar as it points to limitations in research, studies should be designed and *further on point research should be supported* to address the gaps in the literature. It is important to underscore that the lack of evidence of what is *suspected to be missing in the story* (of "what 'the experience' of having a child with Down's syndrome is like") not be taken itself as evidence.

of their preferred reproductive projects. Rather, the HFEA licensing of PGD for Down's syndrome (according to the standards established by the HFE Act 1990 (as amended 2008)), sends the message that this is a "serious condition"[121:s.10.6;144:Sch. 2, s1ZA(2)(a-b)] that carries with it a significant risk of imposing grave suffering (or other compromising features for quality of life) for the affected, prospective offspring and the families of these affected offspring. Given the evidence available, judging Down's syndrome to be a "serious genetic condition" on these terms and for which PGD testing is deemed in principle appropriate, risks propounding a misconception of what life is really like for persons with Down's syndrome and their families. This negative attitude to Down's syndrome is now strongly entrenched within prevailing social norms, both in the UK and throughout much of the globe.[227;443] The argument being put forth here is not so much that allowing PGD for Down's syndrome is wholly responsible for creating the impression that Down's syndrome is an inherently negative genetic condition; rather, allowing PGD for Down's syndrome under the auspices of the present HFE Act and relevant system of guidance in the Code of Practice, wrongfully lends credibility to this impression, thus abetting forces of prejudice, stigma, and unjust discrimination against persons with Down's syndrome and/or their families. The problem here is not whether parents' preferences not to have a child with Down's syndrome are justified or not; the problem is the misuse of authority by the HFEA to underwrite these choices as being based on "something more" than individual or social preferences.[cvi]

This points to wider moral liabilities in a regulatory regime that may (perhaps only inadvertently) work to hide the reality that the reasons for choosing or not choosing to use PGD to prevent having a child with Down's syndrome, have (for the most part) little else to be based upon than personal or social preferences. But, as outlined above, having a child with Down's syndrome is not necessarily regrettable—in fact, it's not even likely to be regrettable. This is not to deny that a life with Down's syndrome—as also for a life with a family member who has Down's syndrome—forecloses certain opportunities. To repeat: some of the disadvantages accruing to persons with Down's syndrome and their families stem from impairment-induced disabilities that social supports may prove practically powerless to redress. The point here is simply to say that most of what is disabling in most cases of Down's syndrome is remediable, and there needs to be much more public debate over how *we as a society will choose, or not choose,* to practically address these needs. In lieu of this, we are allowing what are probably

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[cvi] It may be less harmful for these decisions to be protected as (ultimately) a matter for parental discretion (on the basis of their own reasons to themselves for doing so) rather than being regulated under the current instruments applied by way of UK law and policy.

social preferences to drive regulatory decisions that underwrite restrictive access to PGD testing for Down's syndrome, and *ex post facto* giving ourselves a comforting moral story about "serious genetic conditions" that makes it seem as if that which is "serious" about the associated disabilities is much more determined and life-compromising than is actually the case—neither for the directly affected individuals, nor for their families.

Hence, by allowing access to PGD testing for only "serious" genetic conditions and labelling Down's syndrome as one such condition, in effect the current regulatory system, in this instance, risks participating in a form of social discrimination, possibly compounding forces of stigma, prejudice, ignorance, and general misinformation about the capacities and quality of life possible for person's living with Down's syndrome and their families. In arguing that PGD for Down's syndrome be considered inappropriate under the current regulatory instruments set out in the amended HFE Act as well as the relevant guidance provided in the Code of Practice (8th edition), I do not mean to imply that we ought therefore to disallow access to PGD services for Down's syndrome. But if we do continue to use PGD for Down's syndrome, we should be clear that we cannot honestly think to do so because Down's syndrome is an inherently debilitating, "serious" condition that is, strictly speaking, medically indicated.

Reasons to expunge the list and to resist differential standards

Before moving onto the final section of this chapter, we need to take stock of what has been established. Having referenced the UK legislative standards for licensing PGD as well as the HFEA's guidelines for assessing the appropriateness of PGD applications, it would seem according to our analysis in chapter 2 that the justifications for offering PGD testing for Down's syndrome is in strong tension with, if not contra-indicated by, the relevant evidence given in the on point, scholarly literature. Since Down's syndrome does not seem to pass the HFEA's own tests for determining an appropriate application for PGD, then it stands to reason that it ought not to be treated as a "serious" condition and having it on the list of permissible applications for PGD is not warranted—indeed, it is misleading. Given the current strictures for the regulation of PGD in the UK, de-listing Down's syndrome would seem in order,[cvii] and doing so could go some distance to dampening criticisms of implicit State endorsement for the idea

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[cvii] To repeat: in putting forward this conclusion I in no way would want the reader to infer from this that I mean to question the motives or the "reasonableness" of those parents wishing to avoid having a child with Down's syndrome. The process of de-listing Down's syndrome is put forward here *not* as a move that is in and of itself justified: instead, it is a move that is required *if* the current standards for regulating PGD in the UK are to be consistently applied.

that parents would reasonably wish to avoid having a child with Down's syndrome, and that the reasonableness of such a wish has an objective basis over and above any personal or social reasons.

De-listing Down's syndrome *may be* one step in fending off forces of oppression against this population, but is this just a partial measure for redressing only a narrow set of social inequalities implied by this approach to regulation? There may be more at stake here than just the misapplication of the regulatory instruments for the case of Down's syndrome. After all, just as harms against specific women may not harm only those individuals directly victimized but also harm (directly or indirectly) women as a social group,[90;106:83] so also harms against persons with Down's syndrome risk negatively affecting other relevantly associated social groups of persons living with disabilities.[cviii] I do not mean to hereby suggest that we can simply generalize the results of this investigation into PGD for Down's syndrome to conclude that the same harms implied by having Down's syndrome on this list are of a piece with all other listed conditions on the list. Rather, the point of investigating the case of Down's syndrome is meant to point up the structural blind spots with this approach to PGD regulation. As Cheshire Calhoun, Marilyn Frye, and Susan Sherwin point out, the importance for feminist assessments of harm against disadvantaged social groups needs to be alive to how these are contextualized in *interlocking* systems of oppressive practices.[100:88]

Hence, the case of oppression against the social group of persons living with Down's syndrome in the UK should give us pause to consider what other "disabling" conditions may already be on the list, or will make their way in the future onto the list, without warrant. For instance, the HFEA has granted a license to use PGD to test for autism "at least" once "in the past".[487:pt.7] As such, autism would have to have been considered a "serious" genetic condition at the time of this decision for the license to have been legal in the UK. Nonetheless, as reported by

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[cviii] This point touches on concerns of ableism—the systemic disadvantages rooted in unfair discrimination and unjust inequalities as experienced by those who live in society with various disabilities. Susan Sherwin explains that: "Many feminists understand inequality to be a problem between social groups, not just the individual members of each group. They see systematic patterns of discrimination affecting many different dimensions of society—from the private, domestic sphere to the economic sphere, the world of law and politics, religion and even academic research. … These power differentials can only be appreciated if we see them as affecting social groups, not just the individual members who make up a social group. For any particular woman, it is usually possible to find a man who is more disadvantaged than she—in most cases, because he belongs to another disadvantaged social group (e.g. a racial minority). What is problematic is how women (racial minorities, people with disabilities, non-heterosexuals) generally are likely to be disadvantaged relative to comparable men (or members of relevant privileged groups)."[106:83] From a feminist perspective we need to be concerned about the social injustices for affected social groups, not just individual members of those social groups. See Sherwin[98;106] as well as Young.[90]

the HFEA in a policy briefing note, "many have pointed to this as an aberrant decision."[487] That same 2003 briefing noted that: "In theory the nature of the symptoms is a matter of indifference, although PGD has tended to be licensed only for overtly physical disorders and severe mental retardation. Its extension to disorders formerly classified as 'psychiatric' may be seen as an exceptional step."[487:s.7] Yet, 10 years later (at the time of writing), autism spectrum disorder is, nonetheless, named on the HFEA's website as a listed "PGD condition awaiting consideration."[64] Interestingly, evidence of the kind brought forward in the last chapter is also forthcoming for persons living with autism—some of whom are resistant to the pathologization[488-492], or over-pathologization of their condition[493;494]—and families with autistic members.[495;496] In the emergent neurodiversity community, members with autism are embracing a life with impairments and experiencing them as not necessarily disabling or uniquely burdensome,[cix] even if sometimes very much disadvantaging in society.[493;498] In a report on narratives published on the internet by parents of children with autism, Fleischman observes that "[t]he quality of the child's and parent's life is not viewed as poor."[499:40] The posted stories that were analyzed in this study share a common view of parenting autistic children as being a ‘‘challenge’’; however, Fleischmann also observes that though the parents viewed their experiences of raising an autistic child as non-normal, they did not view themselves as "victims" in any way.[499] Admittedly, this is not proof of the neutrality of traits commonly associated with autism, but it does provide some grounds for scepticism for why it should be considered a "serious"[see 493] condition for the HFEA.

These findings concerning the quality of life potentials (both for persons born with impairments and for their supporting families) weakens the presumptively stronger basis for offering PGD for genetic impairments than for any other genetically detectable human trait or variation. As disabilities scholar and legal academic David Wasserman notes, these findings pose a challenge to a longstanding tradition of making an exception for genetic impairments from bans on other kinds of genetic testing.[500:308;see 501] Asch and Wasserman further warn against the potential harm in continuing to disallow access to prenatal testing technologies but for a selective list of genetic conditions in that doing so would probably only serve to deepen the view that impairments in and of themselves are uniquely burdensome for life. [477;500] As they propose:

[cix] See Karpin and Kristin for a "conceptualization of disability as a distinct way of being in the world, rather than a diminished state of being".[497:29] See also section 1.2 of their book on the question, "Is Disability Necessarily Negative?"[497:21-36]

> …to assume that most genetically detectable disabilities impair the prospects for individual and family flourishing in a way that the other potentially detectable characteristics do not is truly to stigmatize disability. While such stigmatization is understandable when it is displayed by anxious couples awaiting a life-transforming event, it should not guide the public funding of reproductive research or the formulation of reproductive policy.[477]

Alternatively, to better avoid the discrimination and stigmatization of what it means to live with impairments, it may be better to allow testing for every detectable genetic condition according to the dictates of parental choice alone[cx] as the sole basis for embryo selection rather than restricting testing to (what are termed) "serious conditions." Such a permissive[cxi] approach does contain risks of degrading the parental role;[505] at the same time, from a critical disabilities perspective, there are good reasons not to make an exception by allowing testing for disease and impairments but disallowing testing for preferred—or some might say, "desirable"—characteristics. This sort of exceptionalism, so argue Wasserman and Asch, will only serve to entrench the view that impairments are uniquely burdensome.[477;478;500;506] Moreover, this practice of discrimination (built into the HFEA regulatory system for PGD) is difficult to justify in

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[cx] It is an interesting question whether there should be any justifications relevantly required for accessing PGD services over and above considerations involved with factor (a1)[121:s.10.7] (outlined above in chapter 2), namely the views of prospective parents seeking the treatment. Some have argued that where the prospective child would have a fair chance for a "life worth living" despite the impairments associated with a given genetic condition, there are no grounds over and above parental preferences for restricting access to genetic information and the use of PGD or PND to select against affected embryos or foetuses any more so than in cases with "normal" embryos or foetuses. However, in cases where the prospective child would not have a fair chance at a "life worth living," then selecting against the relevant embryos or foetuses might have a stronger justification in that the technologies could be used to prevent undue suffering in a child. The judgment of a "life not worth living," though, is arguably very, very rare (though it has been invoked in cases of wrongful life lawsuits). This line of argument would also stand to challenge the criteria in the Abortion Act 1967 (c.87) (amended by the HFE Act 1990) for selective termination of pregnancy for foetal abnormality (s.37(1)(d)) mentioned in chapter 2. These are important issues here but beyond the scope of this paper. For relevant discussions see Asch;[478] Parens and Asch;[171] Sheldon and Wilkinson;[413] and Wasserman.[500] I thank an anonymous reviewer from *Medical Law Review* for this point and much of its wording here.

[cxi] See Gavaghan,[502-504] who is a strong critic of the current UK regulatory framework. Gavaghan defends a pro-choice position in terms of client access to PGD, and he places the burden of proof on the state to justify restrictive regulation in these matters which would over-ride parents' prerogatives to choose their children. Gavaghan, though critical of the "expressivist argument" made by disability rights activists, argues that an exclusive list of licensable conditions for PGD is still wrongfully harmful to persons with disabilities. Gavaghan argues that reproductive freedom is best served if the state or society regulates in a way that does not betray any value judgments on what kinds of lives are worth preventing, excepting cases of wrongful birth.[502-504]

the face of a rising body of evidence showing the contrary from research on families with children with impairments.[341-348;355;370;477]

In this regard, not limiting PGD testing to what are considered "serious" conditions, and instead allowing testing for impairments as on par with any other detectable genetic traits, could work to dull certain stigmas attached to disabilities linked to the impairments in question. Asch and Wasserman applaud the fact that this would place more responsibility for the resulting decisions in the hands of individual parents—those who ultimately have to raise the resulting children—and take it away from society, in the form of its insurance brokers, hospital boards, ART clinicians, or official regulators like the HFEA who are often charged with, or simply assume, the role of gatekeepers of this technology[478:339;500:309]

But though this argument seems logically compelling, as noted with Kymlicka's assessment in chapter one, most commonly public disagreements over new ARTs are of the "slippery slope variety".[77:254-255] In this case, the slope that is slippery might be psychological rather than logical. Those who might take issue with the proposal just given by Asch and Wasserman might concede that strictly speaking, it is logically possible that making PGD universally accessible across all testable traits could technically lead to further diversity in the human species;[cxii] however, without any social controls other than the marketplace, this seems unlikely and the results of the further commodification of human reproduction might just as well produce more uniformity and less tolerance for diversity in society. For instance, in societies where universal healthcare is the model, social and health supports could wane for those parents who choose to forego PGD and instead birth children whose well-being requires resources that are atypical. In this scenario, there could be public backlash in response to the perception that such lives are preventable with PGD. This could happen in regimes that upheld limited access to PGD (say, on the basis of "seriousness"); this could also happen in those regimes with unlimited access to PGD, but then the consequences could be greater (at least, numbers wise) in that more conditions might be seen as preventable. Indeed, the negative consequences in the latter case could be greater, but need not be so. As Wasserman rightly points out, line-drawing might lull us into believing that at least only those conditions above the threshold of "seriousness" stand to be stigmatized. However, those conditions that fall outside the threshold for

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[cxii] I thank Susan Sherwin for bringing this point to my attention.

"seriousness" need not be immune to stigma. Given that line-drawing tends to entrench the view that impairments are uniquely burdensome, in light of this, public perception might construe those conditions as falling outside the threshold as enjoying this status by virtue of policy processes that involve balancing or compromise, not as the result of "substantially changed beliefs about the burden[s] they impose"[500:301] (Wasserman 2003, p. 301).

Admittedly there are liabilities to a regime that does not impose restrictions on client access, especially in terms of widening the scope for consumerism in matters of human reproduction,[see 507;508] even though more on point empirical research is needed to check these speculative worries. After all, getting rid of state controls for PGD access might obviate the problems of state-driven discrimination. But replacing these with, legally-speaking, open access policies for consumers might leave open other risks for social discrimination against relevant disadvantaged social groups now coming from the effects of the market or through clinicians controlling access to PGD. I have argued elsewhere[117] that this negative potential (that could be exacerbated by moving away from line-drawing [according to the criterion of "seriousness"] as a model for regulating PGD) could be offset, in part, by making sure the availability of this technology is accompanied by relevant educational programming and social supports for the vulnerable that are to be affected: most notably women, children, and persons with disabilities—all marginalized parties, we might remember, whose interests, according to Kymlicka, were either overlooked, or underestimated, by the Warnock Report.

Summary of chapter conclusions

To summarize: in this chapter I have endeavoured to critique some of the structural weaknesses in the UK system of regulating PGD according to which all uses of the technology are prohibited except for applications used to screen embryos for conditions given on a list of (what are considered) "in principle", "serious" conditions. My argument against the principlist approach to assessing what is debilitating about Down's syndrome according to the HFEA's criterion of "seriousness" is that it tends to place the emphasis on the condition itself rather than looking to situational or environmental factors, thus overlooking the complexity of the involved contingencies of associated disability for a life with Down's syndrome. Given the evidence put forth in chapter 2, having Down's syndrome as one such listed, "in principle", "serious" genetic condition is a misrepresentation of the capacities and quality of life possible for persons living with Down's syndrome. This being so, Down's syndrome should be removed from said list. But another option for de-listing Down's syndrome would be to expunge the list altogether. [cxiii] Some of the reasons against having this list include that it is premised on a reified conception of disability that flies in the face of a growing body of evidence that not all lives with impairments are necessarily disabling and that impairments are not uniquely burdensome. Line-drawing efforts (with the power of law and backing of medical authority) that endorse testing and selecting against an exclusive list of genetic conditions while barring prospective parents from selecting for or against other traits, run a significant risk of exacerbating discrimination and stigmatization of persons with the "test-worthy" conditions in question.[478:339;509:4]

Conclusion

The purpose of this thesis has been to query the legitimacy of offering PGD testing against Down's syndrome on the basis of UK law and policies.

The Warnock commission warned of societal dangers with the new reproductive technologies and therefore advocated for legal restrictions on their use. In consequence, PGD in the UK is treated as a legally prohibited activity except for a special set of applications licensed by the HFEA. In this regard, licensed PGD applications are granted for the testing of *ex utero* embryos at "significant risk" for developing into children with inherited, "serious conditions."

Downs' syndrome is considered a "serious condition": it is a licensed application for PGD in the UK, and widely accepted by the public and professionals alike as "serious." Even though the criterion of serious is nowhere defined in UK law and policy, the HFEA's Code of Practice lists several factors for assessing the appropriateness of PGD applications. Drawing upon a wide range of relevant evidence from the scholarly literature, it would seem that a positive judgment of appropriateness of PGD for Down's syndrome as referenced against the factors given in the Code of Practice would be very problematic, if not unjustified. In sum, the impairments associated with Down's syndrome can prove debilitating, but much of what makes these impairments debilitating is remediable through social, medical, and institutional interventions

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[cxiii] Most discussions in the scholarly literature of how to regulate PGD to this point have only looked at issues of client access. There are other unexplored avenues for applying regulation that have received scant attention in the literature and are deserving of ethical, legal, political, and social scientific research. These would include regulation directed more "upstream" in terms of PGD and IVF research and development. In all likelihood, these are important drivers for determining some of the feared outcomes of unregulated access.

or reforms. What is more, much of that which is debilitating about Down's syndrome is in fact the product of social and cultural disadvantages.

As following from a power-focussed *feminist* approach to bioethics, this thesis has endeavoured to take seriously the role that health law and public policy assume in relation to general structures of oppression in society for persons living with disabilities. This means that in the context of PGD, regulation affecting clinical access needs to be critically examined not only in terms of the effects on those individuals directly involved—namely, AHR clients and AHR professionals; it must also be assessed with respect to the surrounding interlocking patterns of bias, prejudice, and unfair discrimination against relevantly affected social groups.[98;106]

Within the current structures for regulating PGD in the UK according to an exclusive list of "in principle", "serious"-and-therefore-licensable conditions, state and medical authority is, in effect, underwriting those reproductive choices to prevent the birth of persons with Down's syndrome as being justified for reasons over and above their actual status as expressions of individual or social preferences. With the maintenance of this list, the state is in effect showing more support for, and conferring special legitimacy on, those choices to genetically screen against having children with Down's syndrome as compared with the choices to resist this well-established, widespread social trend. The danger of this imprimatur (as given by state, legal, and medical authority) is that it feeds historically oppressive forces of unfair discrimination against this disadvantaged social group and reifies false beliefs about the quality of life potential of persons with Down's syndrome and their families.

One response at this juncture would be to de-list Down's syndrome and leave in place the other conditions on the HFEA's list. But is the case of Down's syndrome so particular that taking it off the list would be an adequate response to the criticisms levelled above against the UK's regulatory law and policies pertaining to PGD. I suggest that the case of Down's syndrome is a telling one, and indicative of wider structural problems with the current principlist approach to regulation according to (vague) determinations of "seriousness." In effect, this approach places the emphasis for explaining disability on the condition itself rather than looking to situational and environmental factors, thereby ignoring the complexity of the involved contingencies of associated disability for a prospective life with impairments. Another way to de-list Down's syndrome would be to expunge the list altogether.[cxiv] Doing so would go some distance towards

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[cxiv] Another, more conservative approach, might argue that we could keep the HFEA's list of licensable, "serious" conditions, and address the risks of negative messaging through anti-stigma education. But this

avoiding some of the harms we would expect other relevantly similar social groups of persons with disabilities to experience as a result of carrying a disease label of "serious" as a function of being named on this discriminatory list.[cxv]

The Warnock Committee contended that how we seek to control the new reproductive technologies reveals various moral ideals for what kind of society we wish to protect and promote (as expressed in and through law[see 81]). In his analysis of the Warnock Report, Kymlicka has argued that taking morality seriously in the design of laws and public policies to regulate new ARTs requires us (at least) to i. identify which people are affected by new ARTs; and, ii. ensure "that [new ARTs] are used in such a way as to promote, or at any rate not to harm, their legitimate interests."[77:251] It has been an overall burden of this thesis to show some of the main problems with the on point UK law and policy instruments for regulating PGD in terms of their built-in negative potential for harms against persons living with disabilities.

By providing the groundwork for regulating new ARTs and recommending where barriers to its uses should be placed, the Warnock Committee owned that not only were they engaging the question of "what kind of society can we praise and admire", but also "[i]n what kind of society can we live with our conscience clear?"[61:sec.8] If the current HFEA model of regulation for PGD can ethically fail the above-mentioned populations of persons living with disabilities in ways that actually serve to reproduce forces of oppression germane to the "kind of society" that we should not rightfully hope to (continue to) live in "with our conscience clear",[61:sec.8] then perhaps this argues for a reassessment of some of the built-in assumptions and starting points of the UK licensing programme for PGD. What is given here in this thesis is meant as a ground clearing exercise (*à la* Locke), in order to start considering the question of how the default protectionist policies on client access to PGD in the UK, unwittingly or not, are at risk for harming social groups of persons living with disabilities. These harms are entrenched in some of the "blind spots" of UK ART regulation that I have tried to consider with this thesis—blind spots that call forth consideration of law and policy alternatives that would be less harmful and more equitable to persons living with Down's syndrome as well as other similarly positioned

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would risk sending a double message rather than a clear one. As Elisabeth Gedge notes: "Official limitations on [PGD] testing suggest that formal authority is required to curb disableist sentiments, a disturbing message in itself".[69:248] Gedge, enlisting Gavaghan's arguments,[510] further explains that if the negative message "is considered to be sent by society in general, …official limitation of PGD will merely deflect it and it will manifest itself in other ways." [69:248] By contrast allowing testing for any traits, whether they are considered disabling or not, would send only the message that society upholds reproductive freedom and would affirm women in retaining control over their reproductive projects.

[cxv] Note that I have not said that that this would be true for all conditions on this list.

disabilities groups. Feminist public policy analyst and ART researcher Abby Lippman reminds us that in developing, distributing, and regulating ARTs, "[t]here are choices to be made and the choices will reflect our values and ideology. How we choose our culture (by the routes we take) is no less problematic than how we choose our children, and consequences from both will be among our legacies."[118] As archaeologists know, future generations can learn about the values and norms of previous generations as much by examining their rubbish[cxvi] as by examining their achievements. Given this analogy, what might our descendants learn about our culture by reflecting on the law and ethics of regulating PGD over the past twenty years in the UK and observing not only which policy choices have been taken up, but also which ones have been overlooked and discarded?

[cxvi] I owe this insight to a former teacher of mine, a Mr. William Schroeder.

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