

Introduction

WHY PERFECTING PREGNANCY?

Over the last twenty years there has been a proliferation of diagnostic technologies aimed at identifying and eliminating potential abnormalities in future children.¹ Alongside this rapid development in diagnostic technologies we can chart an equally rapid development in the technologies of “reproductive management.” These include new tertiary degrees in genetic and prenatal counseling;² the new centrality of the role of the clinical geneticist;³ the inclusion in laws, ethical guidelines,

¹ Prenatal testing technologies have expanded so that we now have on the horizon the potential to take a simple blood test to test for a number of disabilities including Down syndrome: see I. Sample, “Simple Blood Test for Down’s Syndrome Is on Its Way, Say Scientists” *Guardian*, 6 March 2011: see <http://www.guardian.co.uk/science/2011/mar/06/downs-syndrome-simple-test-in-pregnancy> (accessed on 27 June 2011). Preimplantation Genetic Diagnosis (PGD) can now test for more than 150 different disorders: “PGD Conditions Listed by the HFEA”: see <http://www.hfea.gov.uk/cps/hfea/gen/pgd-screening.htm> (accessed on 27 June 2011). Preconception testing is now able to identify more than 400 recessive genes for severe childhood disorders: National Health Service, “Gene Test ‘Predicts 448 Child Diseases’”: see <http://www.nhs.uk/news/2011/01January/Pages/dna-genetic-test-for-parents-before-pregnancy.aspx> (accessed on 27 June 2011).

² See the Human Genetics Society of Australasia, “Guidelines for Training and Certification in Genetic Counselling” 2010 GL/01, 20 February 2010 at 8.

³ See, for instance, the Human Genetics Society of Australasia, “Guidance: Clinical Geneticist’s Role” 2010 GD01, August 2010, which states: “Clinical

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and clinical best practice notes of a requirement for genetic counseling; and a proliferation of online fact sheets and decisional aids. We have also seen a burgeoning of informal networks of information exchange via the Internet, parenting magazines, disability support groups, and the media more broadly. At the same time that this has been happening there has been vigorous public debate in countries all over the world about the broader social implications of these diagnostic technologies. A significant portion of this debate is *not* concerned with the moral or legal status of the fetus or embryo per se but, rather, with the use of these technologies for eugenic and discriminatory purposes.⁴ These debates around new prenatal diagnostic technologies often focus on the question of whether it is *right to choose* “better,” “unaffected,” “nondisabled” children.⁵ While this is a concern of our book, too, our focus is different. We called this book “Perfecting Pregnancy,” rather than “Perfecting Progeny,” for example, because our focus is on the way that these discourses regulate and influence the behavior of people who are already in the world. One of our primary goals in writing this

geneticists see referred patients for diagnosis, management, genetic testing and genetic counselling.”

⁴ The critical literature on this point is expanding: see, for example, E. Parens and A. Asch, “The Disability Rights Critique of Prenatal Genetic Testing: Reflections and Recommendations” (1999) 29(5) Special Supplement *Hastings Center Report* S1; J. Gillott, “Screening for Disability: A Eugenic Pursuit?” (2001) 27 *Journal of Medical Ethics*, Suppl II: ii21.

⁵ R. Scott, *Choosing between Possible Lives: Law and Ethics of Prenatal and Preimplantation Genetic Diagnosis* (Oxford: Hart Publishing, 2007); S. Wilkinson, *Choosing Tomorrow's Children: The Ethics of Selective Reproduction* (Oxford: Oxford University Press, 2010); J. Glover, *Choosing Children: Genes, Disability and Design* (Oxford: Clarendon Press, 2007); L. Skene and J. Thompson (eds.), *The Sorting Society: The Ethics of Genetic Screening and Therapy* (New York: Cambridge University Press, 2008); R.M. Green, *Babies by Design: The Ethics of Genetic Choice* (New Haven, Conn.: Yale University Press, 2008); A.R. Chapman and M.S. Frankel, *Designing Our Descendants: The Promises and Perils of Genetic Modifications* (Baltimore: Johns Hopkins University Press, 2003); M. Sandel, *The Case against Perfection: Ethics in the Age of Genetic Engineering* (Cambridge, Mass.: Harvard University Press, 2009).

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book was, therefore, to find out just who *is* making the decisions about the way in which we (society, women, mothers) manage the potential for disability in our reproductive futures.

In order to answer this question we begin by unpacking some fundamental assumptions about the “badness” of disability and the “realness” of reproductive choice.⁶ Some scholars have argued, for example, that prenatal testing and abortion and/or selective embryo implantation using PGD express a discriminatory or negative attitude toward people who have the disability for which such testing is offered. Concerns have also been raised about whether “disability” has been, or may be, given too broad a construction in these contexts. This theme is a major consideration of our book. Second, others, including us, have written about the pressure on women – from medicine, law, and society – to manage their pregnancies and potential pregnancies to ensure the “best” possible opportunity for a “good,” “healthy,” “normal” outcome, even if this may require the abandonment of the process altogether. Thus we find that women in contemporary Western society are encouraged to imagine their pregnancies as processes that can be perfected, indeed that they have a responsibility to perfect.

In order to understand how this kind of framing of reproductive options is instantiated, we chart the regulatory history of abortion, prenatal testing, and preimplantation genetic diagnosis in a number of jurisdictions including Australia, the United Kingdom, the United States, Canada, and some European countries. We examine how law has regulated, and continues to regulate, these fields but also, perhaps more important, how it *does not*. What we discover is that medical

⁶ See, for example: T. Shakespeare, “Debating Disability” (2008) 34 *Journal of Medical Ethics* 11; S. Edwards, “The Impairment/Disability Distinction: A Response to Shakespeare” (2008) 34 *Journal of Medical Ethics* 26; T. Koch, “Is Tom Shakespeare Disabled?” (2008) 34 *Journal of Medical Ethics* 18; R. Gillon, “Is there a ‘New Ethics of Abortion?’” (2001) 27 *Journal of Medical Ethics* 5; J. Savulescu, “Is Current Practice around the Termination of Pregnancy Eugenic and Discriminatory?” (2001) 27 *Journal of Medical Ethics* 165.

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professionals play a critical role in determining what kind of diagnostic testing is made available, who may have access to it, and the reasons that will justify the grant of such access. This is despite the fact that many countries (Australia, the United Kingdom, New Zealand, and a number of European countries) have responded to innovations in diagnostic technologies in the area of reproduction with a period of intense regulatory activity.

Toward the end of our study we begin to chart the potential impact of new technologies that are being developed, including inheritable genetic modification techniques and – perhaps most important – the proliferation of preconception technologies that, paradoxically, push our reproductive futures backward to an earlier point in time so that actions taken yesterday and today are conflated with the child possibly conceived tomorrow.

In the face of these novel technologies, the overwhelming question for regulators has been whether and, if so, how to limit reproductive decision making. One particularly popular strategy worldwide has been to limit the use of such technologies to those circumstances where the aim is to avoid “serious disability.” However, while concepts of seriousness, disability, and of course normality are currently used to frame legislative limits, they are, more often than not, left undefined.

In *Perfecting Pregnancy* we examine this use of “serious disability” as a tipping point in the regulation of prenatal testing (PNT) and abortion and preimplantation genetic diagnosis (PGD). We ask whether this concept can, or should, do the work required of it by legal and ethical regulatory frameworks.

APPROACH AND ANALYSIS

A crucial dimension to our approach is our exploration of the meaning of “serious disability” as a legal concept compared with a medical concept. By drawing upon legal texts, policy documents, and the

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growing empirical literature about the attitudes and experiences of those engaged or potentially engaged by these reproductive technologies and their understandings of disability, we examine whether there is a shared normative framework that grounds legal conceptualizations. In this book we focus on the attitudes of regulators as well as those of health care professionals, including genetic counselors, clinical geneticists, in vitro fertilization (IVF) practitioners, and others who work in the industry. However, we are also engaged in an ongoing study that explores the attitudes of women and patients to these technologies and the conceptualization of disability. The material from that research will be the subject of subsequent publications.

In this book we have drawn heavily on existing empirical literature and draw fresh insights from our own empirical research, in which we interviewed Australian regulators and clinicians. These interviews are part of an ongoing qualitative study of expert accounts of the use of preimplantation genetic diagnosis and prenatal testing technologies in Australia.⁷ Given the small number of participants, we include these excerpts for their anecdotal value. They offer individual insights that may not otherwise have been accessible. Participants so far included have been from the federal sphere and the states of New South Wales, Victoria, South Australia, and Western Australia. We have included excerpts from 12 interviews conducted between June 2010 and October 2011 in this book. The interviewees include regulators, clinical geneticists and genetic counselors. In each case the experts were selected because they were known to be key actors in the field. The broad intention of our interviews was to elicit participants' views on how the meaning of "serious disability" is determined in different clinical and regulatory contexts and by whom. We also wanted to find out what role the law plays in shaping this meaning.

⁷ This work has been undertaken as part of Australian Research Council Grant DP0988103, "The Legal Function of Serious Disability in Prenatal and Neonatal Healthcare Settings." Ethics approval was obtained from the Sydney University Ethics Committee and the University of Technology, Sydney, Ethics Committee.

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Consequently our research approach was intended to open a space for discussion and to allow for the flexibility to adapt the interview depending on the direction the interviewee's response dictated. It was specifically intended to be iterative. Thus, as will be seen in our discussion of these interviews in later chapters, we adopted a "narrative-based" research methodology. Our interviews were conducted as "guided conversations."⁸ Open-ended questions were used with a set of prompts so that all participants were asked a select set of common questions. All expert participants were contacted by letter/email and asked whether they would be interested in providing their views. Each participant was then interviewed in person for 45 minutes to an hour. The interview was recorded and transcribed, and participants were given the opportunity to review the transcripts and amend them where they felt necessary. All contributions were provided on condition of anonymity and interviewees have been given numbers. Clinicians are represented by a *C* and regulators by an *R*. Chapters 2, 4, and 5 contain excerpts from these interviews.

THE STRUCTURE OF THE BOOK

Framing Concepts – Disability and Risk

Disability and its avoidance is a concept that frames clinical approaches to the routine management of pregnancy, public health prenatal screening programs, the development of new advanced techniques such as PGD, women's experiences of pregnancy, and legal and regulatory efforts to delimit the range of appropriate responses to prenatal testing and diagnoses. Chapter 1 examines the use of disability as a framing concept for

⁸ J. Lofland and L.H. Lofland, *Analyzing Social Settings: A Guide to Qualitative Observation and Analysis* (Belmont, Calif.: Wadsworth, 1984); K. Ehrich, C. Williams, and B. Farsides, "The Embryo as Moral Work Object: PGD/IVF Staff Views and Experiences" (2008) 30(5) *Sociology of Health & Illness* 772.

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reproductive decision making and asks: “What does disability mean?” “is it always bad?” “how is it understood and by whom?” and “what are its particular dimensions in the context of prenatal screening and selection practices?” We begin the chapter by noting that, despite the heavy reliance on the concept of disability to frame both legal and medical responses to diagnostic testing technologies, disability remains a somewhat enigmatic concept. We follow this observation with a thorough interrogation of the varied and rich scholarship exploring the question of what disability is and how it should be understood. We consider a number of significant debates concerning the way in which disability is constructed in contemporary society, and we unpack some of the key insights produced by the disability studies movement. In the context of prenatal testing we note the argument sometimes made by disability rights scholars that once an abnormality is detected, women do not have a true choice about whether to continue the pregnancy because a diagnosis of disability is portrayed unfavorably. At the same time, however, it is recognized that women do want to be informed and to be able to make choices freely about whom and how they reproduce. In addition to this, we note that there is a strong feminist concern that prenatal testing is presented to women as being something they have a responsibility to undergo. Although the literature provided to pregnant women about prenatal testing is at pains to present as *genuine* the choice confronting women about whether to (a) accept the offer of testing and (b) have an abortion for an abnormality if identified (or in the case of PGD to select against disability), in fact it sometimes creates an environment in which pressure is placed on women to make particular decisions to reproduce “responsibly.”

In Chapter 2 we turn to a discussion of how the debate about the construction of disability is informed by (and/or informs) the discourse of risk management that pervades prenatal screening and testing. We explore how risk, as a conceptual apparatus, is deployed in legal and biomedical discourses, government and policy discourses, cultural and media discourses, and, of course, the minds and bodies of individuals. Chapter 2 enlarges upon the idea that social attitudes and pressures limit

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women's freedom to make reproductive decisions for themselves. The argument here is that we live in a "risk culture": that is, a culture in which the concept of risk has become a pervasive conceptual tool driving regulatory responses and disciplining pregnant women. As feminist legal theorists, in this chapter, we explore the way risk discourse gives rise to both legal and nonlegal forms of regulation enacted on and through the bodies of pregnant and potentially pregnant women. This is done while recognizing the tension between the opportunities risk discourse offers for informed reproductive decision making and the burdens it imposes.

A Genealogy of "Serious Disability" as a Regulatory Concept

Although concepts of disability and normality are currently used to frame legislative limits, they are, more often than not, left undefined. In Chapters 3 and 4 we argue that in order to understand how this regulatory approach has evolved, it is important to consider the context. Early regulation of PNT and abortion was shaped by the background events of the rubella/thalidomide outbreaks while regulation of PGD took place in the context of the project to map the entire human genome and the developments in cloning technology. In Chapter 3 we trace the development of abortion law reform in Australia and the United Kingdom over the period of the last 50 years, paying particular attention to the way in which abortion to avoid "serious handicap" emerged in law as a distinct category of legal abortion. Although this reform has not been uniformly enacted, it nevertheless provides a conceptual apparatus that takes on a wider relevance. In jurisdictions that do not have such a provision in law, decisions about late termination are nonetheless made by reference to this concept by clinicians acting within their professional guidelines. Furthermore, "serious disability" laid the conceptual foundation for legislative intervention into the field of PGD. Thus, in Chapter 4 we examine the history of regulation of

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preimplantation genetic diagnosis over the last 20 years in Australia and the United Kingdom. In both chapters, we also map out the contemporary legal terrain in Australia, the United Kingdom, Canada, the United States, and a number of European countries. In both the PGD and PNT contexts, most of the legislatures discussed have ultimately devolved responsibility for determining when disability avoidance technologies can be used to licensing bodies or to the clinicians themselves. Where clinicians must seek approval from a licensing body that has been given the responsibility of making such determinations, more often than not that body draws on clinicians' expert guidance to assess whether treatment meets the relevant criteria. In the absence of a licensing body and any other decisive legislative guidance, the decision lies in the hands of the individuals seeking treatment, with the medical profession acting as a "gatekeeper." Despite there being some concern about the appropriateness of reposing in the medical profession this gatekeeping function, as we see in Chapters 3 and 4, the overall pattern of regulation has nonetheless been to favor this approach. In both the abortion and PGD contexts, the principal legislative means to restrict the medical profession's discretion has been by inserting the qualifying term "serious" (or some version thereof) to limit the kinds of disabilities these techniques can be used to avoid. In Chapter 4 we draw on interviews with regulators who discuss whether the absence of a legislative or regulatory definition for this qualifying term is appropriate. They consider whether it is meaningful as a legal restriction on medical decisionmaking insofar as it creates a framework for decisionmaking that requires, at the very least, discussion and forethought before a determination is made.

The Future for Existing and Emerging Technologies for Avoiding Serious Disability

Although legislators have resisted providing a definition of "serious" handicap, disability, or condition, it remains the case that the term

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must be given meaning in order for the law to function. In the context of prenatal testing and abortion, the available evidence suggests that doctors in Australia and the United Kingdom hold quite divergent views about which conditions are seriously disabling. In Chapter 5, we provide a closer examination of the various strategies – both substantive and procedural – that have been used to structure or guide the decisionmaking of individual clinicians as they interpret the meaning of “serious disability” in the abortion context, and we compare these with regulatory approaches to the meaning of disability and serious disability in the field of PGD.

We then turn to the question of how decisions about “serious disability” are made by women, their partners, and clinicians through an analysis of the available empirical research and our discussions with relevant clinicians. We observe that often decisions about whether a disability is “serious” are complex and contextual. Accordingly, the question of what constitutes a seriously disabling condition cannot be answered simply by focusing on the potential child’s prognosis. Although the perceived seriousness of the condition in question is an important consideration, a number of other factors and pressures bear upon decision making across these two fields. These include the attitudes, experiences, and resources of the family into which the child would be born; the capacity of technology to detect abnormalities in utero or in vitro; the professional regulation of clinician discretion within clinics and hospitals; and the moral and social status of embryos and fetuses.

A key point to emerge from our analysis is that the interpretation of “serious disability” is informed by social and moral perceptions of the avoidance strategy in question. We therefore consider what the implications might be for law and clinical practice. If there are material differences in the perception of moral or social risk associated with abortion compared with PGD, a further question that arises is whether alignment of the interpretation of “serious disability” in the two fields will be sustainable into the future. We begin an exploration of this

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important question through our interviews with clinicians involved in the provision of PGD and prenatal testing services.

In Chapter 6 we explore technologies that are currently under development that may create new dilemmas for legislatures and ethicists. While the federal and state governments discussed in this book seem to be ambivalent about imposing constraints on reproductive decision making to avoid disabilities (however they may be defined) through PGD and selection or PND and termination, there have been limits placed on other kinds of reproductive technologies such as cloning and inheritable genetic modification. Here we take an extended look at some new technologies that are likely to be made available for clinical use in the near future, and we ask whether they pose new or different regulatory questions. We begin by exploring recent developments in prenatal testing that have led to the creation of noninvasive blood tests early in pregnancy for a range of congenital disorders previously only diagnosable at the 15- to 20-week stage by chorionic villus sampling (CVS) or amniocentesis. This new simpler and faster test may eventually allow clinicians to do a full fetal deoxyribonucleic acid (DNA) scan. Once this is possible we may find that the range of what constitutes a normal unaffected embryo is narrowed. Consequently this technology has the potential to change our understanding of disability, pregnancy, and normalcy.

In the second half of the chapter we examine the potential for disability avoidance using gene manipulation therapies. Gene therapy has been argued by some to be the next obvious stage in disability avoidance technology. When conducted on an embryo, gene therapy can lead to an inheritable modification. Inheritable genetic modification is illegal in almost all of the jurisdictions that we examine in this book, and yet it offers the possibility of treating rather than avoiding disability. In other words, the affected embryo or fetus (or gamete) can continue to develop after the particular abnormality or defect has been treated. It does not, however, resolve concerns about eugenic impulses. Genetic modification inevitably involves a preference for some traits

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and the rejection of others. We focus on one genetic manipulation therapy in particular – that used to avoid mitochondrial disease. Chapter 6 concludes with a brief examination of advances in preconception testing technology aimed at identifying carrier status for severe childhood disorders in adults. We consider the implications of this kind of testing, which occurs prior to the existence of even a conceptus, and speculate on how it constitutes the ultimate demand for responsible reproduction toward a perfected pregnancy.

We conclude the book by answering a series of questions that we have considered throughout. First, how do we understand the concepts of “disability” and “normality” when used as threshold categories for framing regulation? Second, can we offer a feminist reading of the critical disability studies critique of PND and PGD without ceding that these two positions are incompatible with one another? Third, drawing on the burgeoning field of empirical texts that examine attitudes and responses to both disability and prenatal and preimplantation testing technologies, we ask how the concepts disability and seriousness are understood both within and without the law. Finally, we combine these empirical and theoretical elements to develop a framework, from a legal/regulatory perspective, for thinking through the challenges of disability avoidance technologies and the pursuit of perfection on and through the bodies of women.