Prenatal and Preimplantation Diagnosis

Joann Paley Galst • Marion S. Verp
Editors

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The Burden of Choice

Springer

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This book is dedicated to my 3-Ds who have influenced my life in immeasurable ways.

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To my children and grandchildren whose love and support have meant everything. May they always have choices.

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Preface

In the past, a woman became pregnant and gave birth to a baby, navigating the pregnancy with little or no information about the fetus and unaware of problems that might be present until after delivery. More recently, thanks to advances in genetics, it has become possible to sequence individual human genomes and detect in a preimplanted embryo or a fetus the presence or absence of not only entire chromosomes but also single nucleotide variations. We have entered an era of rapidly expanding options for noninvasive prenatal screening and testing. Safe, reliable, and inexpensive screening is now available to women early in pregnancy as an alternative to invasive procedures such as amniocentesis and chorionic villus sampling. As a result of the availability of multiple options for prenatal information gathering, women and their partners are increasingly faced with decisions regarding whether to undergo preimplantation or prenatal screening and if so, which methods to choose.

Screening brings with it the possibility of receiving information indicating actual or potential fetal abnormality prior to transfer of an embryo to the uterus, or during an ongoing pregnancy. As prenatal screening becomes more routine not only for women above a particular age but also for all women, parents often perceive it as just another opportunity to confirm that all is well with the pregnancy; they do not necessarily understand that this testing is optional and fraught with downstream implications. This typical parental mindset underscores the need for pre-screening counseling in order to obtain truly informed consent. Post-screening counseling to address the need of some for additional diagnostic testing and to deal with their psychosocial needs must also be available as couples decide how to deal with the possibility or fact of a fetal anomaly.

After receiving information of an anomaly during an ongoing pregnancy, parents are forced to make decisions of enormous difficulty, including that of whether to continue the pregnancy. Complexity is added to these decisions as the severity of the problem is sometimes undeterminable while the fetus is in utero. For most parents this is experienced as a Sophie's Choice decision with no good option. Despite the availability in 2015 of more genetic information than ever before, some of the information still is of uncertain clinical significance, which only exacerbates parental uncertainty and anxiety.

Our purpose in editing this book has been to expand the awareness of professionals from multiple disciplines, including obstetricians, reproductive endocrinologists, clinical geneticists, genetic counselors, and mental health professionals, of the prenatal screening and diagnostic tests available, the information tests can provide as well as their limitations, and the emotional ramifications of prenatal/preimplantation diagnosis, prenatal decision-making, pregnancy interruption for fetal anomaly, multifetal reduction for high-order multifetal pregnancies, and preimplantation choices involving selection of only the "best" embryos. We believe this cross-fertilization among fields to be particularly important in light of the growing use of prenatal diagnostic techniques as well as the expansion of screening to include pregnant women of all ages and genetic backgrounds. It is our hope that professionals with enhanced sensitivity to these emotionally charged and potentially traumatic situations will be in a better position to assist and support patients in their decision-making and in coping after decisions have been made.

We have assembled a group of experts in their fields to inform our readers so they can better address fundamental questions, i.e., what tests are currently available and with what reliability and risks; how clinicians can best assist their patients in weighing the risks and benefits of screening and diagnostic testing; how the patient's values and preferences can be incorporated so the clinician and patient can collaborate in determining her optimal testing strategy; and how we can best support our patients during their decision-making process and after having made their decisions. Collaboration between patient and care giver is vitally important since there is no universal correct answer in prenatal genetic testing, only the answer that is right for each woman. She and her partner, if one is involved, are the ones who must live with the consequences of their decision. This reality presents professionals and their patients with both the justification for patient autonomy and its cost in decisions about pregnancies and pre-pregnancies with an anomaly.

Organization of the Chapters

Part I presents a medical perspective. We begin in Chapter 1 with a discussion by Dr. Verp of the details and recent innovations in the field of prenatal genetic screening and diagnosis, including the multiple new options for preconception carrier screening. Dr. Simpson continues this theme in Chapter 2, reviewing the evolution of preimplantation genetic diagnosis from traditional to novel indications, the unique diagnostic approaches required given the small amount of genetic material available at this stage, and the inherent ethical dilemmas.

Dr. Dungan (Chapter 3) explores the many factors influencing couples' choices about whether to terminate an abnormal pregnancy, and the frequency of termination with different chromosomal and single gene disorders. Differences in termination rates in different countries are also enumerated. In Chapter 4 Dr. Otaño and coauthors delineate the classification of anomalies used by dysmorphologists and geneticists, review ultrasound screening guidelines offered by professional

organizations, and detail management and differential rates of termination for different structural abnormalities.

Dr. Evans and colleagues review in Chapter 5 the historical development of the procedure of fetal reduction, their own groundbreaking contributions as well as data and important contributions made by other centers, and the difficult choices and novel approaches possible. In Chapter 6, Dr. Derbyshire addresses the question of fetal pain, a controversial area much benefiting from his detailed knowledge of the science and his unbiased acknowledgement of the unknowns of this subject.

Dr. Lalor in Chapter 7 presents her research, conducted in a country (Ireland) where termination of pregnancy for fetal anomaly is not allowed, into typical patient assumptions during prenatal screening and diagnosis, and the effect of the unexpected finding of a fetal anomaly on the parents' assumptive world. She stresses the importance of both the language used by professionals in sharing information with their patients and the recognition of individual coping styles that patients use and with which they are most comfortable receiving diagnostic information. She offers guidance for most effectively approaching patients during initial sonograms and in the obstetrician's office when sharing troubling news.

In Part II (Chapter 8), Professor Koch presents an overview of the legal landscape of regulation and oversight of prenatal and preimplantation diagnosis across the United States.

Part III offers alternative social perspectives that may help the reader better understand patient values and preferences as they are factored into their decisions. Dr. Mahowald (Chapter 9) reviews bioethical principles of autonomy, nonmaleficence, beneficence, and justice that can guide individuals in making their decisions, along with an example of difficult decisions in preimplantation genetic diagnosis.

In Chapter 10 Professor Anderson explores the wide range of spiritual and religious beliefs, both across and within religions, on topics related to prenatal and preimplantation diagnosis. Summaries of original texts from the Hindu, Jewish, Islam, Catholic, and post-reformation Christian religions are offered.

The perspective of disability scholars and activists is presented in Chapter 11 by Professor Wasserman. While fully accepting a woman's right to choose, he raises concerns about the use of prenatal testing to select specifically against disabilities, as these decisions may be based on misconceptions that able-bodied people have about life as a disabled person. Dr. Blizzard (Chapter 12) presents a feminist perspective on prenatal and preimplantation diagnostic testing using bioethical concepts of patient autonomy and informed consent as a backdrop, suggesting that the availability of these tests, the information they offer and the subtle suggestions of the type of baby one "should" produce, adds pressure to women in making their reproductive choices.

Part IV offers a psychosocial perspective on making decisions after receiving a diagnosis of fetal abnormality and coping with those decisions. Dr. McCoyd (Chapter 13) suggests that decisions made after a diagnosis of fetal anomaly are highly contextual, driven by factors such as a woman's religious views, beliefs about quality of life, availability biases, access to support networks, and her sense of her ability to cope with the selected option. She stresses the inevitability of grief

for any of the possible decisions that are made and offers strategies to provide care for the woman and her partner after a fetal anomaly is identified.

While women have the right to make choices regarding a pregnancy with a fetal anomaly, these choices, particularly to interrupt a wanted pregnancy, typically have emotional sequelae which can result in traumatic and/or complicated reactions. In Chapter 14 Dr. Galst discusses targeted psychotherapeutic interventions that mental health professionals can offer their patients, both after making their decisions and during a subsequent pregnancy. Dr. Bindeman (Chapter 15) stresses parents' need for support after making a termination decision and, because of the continuing stigma surrounding abortion, the additional difficult decisions regarding disclosure of their traumatic loss, depending on the support they expect to receive. Suggestions for communicating with existing children to help them process the experience of no longer having the expected new baby join their family are also presented.

The final Postscript of the book presents a patient's perspective of a pregnancy termination for fetal anomaly. The experience of being diagnosed with a fetal anomaly and the decision-making incorporating their religious beliefs and expectations as parents are just two of the multiple and complex factors that this author and her partner included in their thoughtful decision-making process. She discusses, as well, the shame she felt, imposed by the polarized political discussion of abortion in the United States, and the happiness she experienced with the birth of a healthy child, capable some day of making his own choices about living a fulfilling life.

Our hope is that this book will contribute to the discussion across multiple professional fields about prenatal testing and diagnosis. The many complex variables involved in patients' decisions about whether to undergo testing and how they deal with the burden of choices during a pregnancy make it clear that we must resist the temptation to assume that we understand what patients will want. Rather, we must listen for their unique values, preferences, coping styles, personal history, and other input regarding these ethical dilemmas, and respond with care and compassion. In this way, we can help these vulnerable patients with truly personalized health care, in hopes that, with time, they are able to find peace with the difficult decisions they have made.

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Joann Paley Galst:

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Marion S. Verp:

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