/BOOK REVIEW

Doubt and Certainty in Genetic Diagnosis

LISA BAUMBACH

As any clinically oriented geneticist already knows, there is often only the finest of dividing lines between "latest research," "clinical protocols," and "currently available techniques." The technical and ethical issues surrounding preimplantation diagnosis are in the news. This can mean that people, including professionals, are frequently semiinformed, armed with just enough information to be dangerous. But every day our clients—the patients, or their parents to be, at least—inquire implicitly, if not explicitly, about the current methods both for detecting and for treating genetic disease. They have the right to know about the options open to them and their implications. We, as clinicians, cannot discharge our responsibilities to them unless we not only provide the fullest of information, but also provide it in a way in which they can use it. That necessitates a broad view of the field, one that is not always compatible with the somewhat telescopic gaze of the specialist. There is a need, therefore, for a book like this one, a book that attempts to weave together a set of otherwise unconnected technical, clinical, legal, and ethical threads.

Sometimes the best sense of where we are going comes from looking back at the tracks we have just made. Robert Edwards, whose clinic at Bourne Hall near Cambridge (U.K.) was responsible for much of the pioneering work in human preimplantation technology, has edited *Preconception and Preimplantation Diagnosis of Human Genetic Disease*. Edwards' close involvement with the recent technical history of the field is clear from the authoritative tone in which the story is told. Writing often in the first person, he strides confidently through his subjects—from the earliest efforts in typing spermatocytes and selective fertilization, through DNA-based prenatal diagnosis, to the technologies of the present day such as the detection of fetal cells in maternal blood.

Not all of the chapter authors have shared, as Robert Edwards has done, their recent experiences. The second section of the book, for instance, looks at the development of prenatal diagnosis and preconception technology. It is inclined to generality, with some discussion of particular applications of preimplantation techniques. Some of the space could, perhaps,

have been better spent in expounding the authors' own experiences—undoubtedly extensive—with the various protocols currently available for preimplantation diagnosis.

Information, for instance, on the number of patients that the authors had seen, the rate of successful pregnancies, and the general problems they faced in diagnosis might have shown concerned practitioners the way to solve some of their own difficulties. They might also have provided information that could be passed on to reassure patients, or at least to give them a more complete picture.

Throughout the book, many chapters have been written with a less-than-clear view of the target audience or, sometimes, of the scope of the book. For instance, chapter 8 describes in great detail a whole raft of DNA-based diagnostic methods and their variants, many of which simply do not apply when all you have to study is a single cell. There is a great deal of space given over to the description of the molecular biology of diagnosis. Much of this material will be thoroughly familiar—and therefore superfluous—to a majority of the book's intended audience, those with a grounding in genetics. At the same time, that same material goes far too deeply into the technical detail for obstetricians.

The final section of the handbook deals with ethical and legal aspects of the diagnosis of human genetic disease. This is an area fraught with uncertainty. There are no straightforward answers. When, precisely, does one become a human being? Who has a right to know about the genetic status of an unborn fetus? What responsibilities attach to the accession and use of that knowledge? Across such terrain, the logic of the law and the guidelines of ethics and morality will rarely run parallel, especially when crossed—as they frequently are—by the thrust of technical advance.

This section sets out the laws on many of the issues. It defines prenatal diagnosis in legal terms. It outlines the social and legal consequences of prenatal diagnosis, current laws on the termination of pregnancy, and on contraception. It defines "parenthood," not always a simple task when dealing with the lives of the unborn.

The book does have a good deal to commend it. The writing style is relaxed and the photographs throughout are excellent, beautiful even. If the book goes into a second edition, however, it will be necessary to define more carefully the intended audience and their information needs. For both obstetricians and geneticists alike, the next edition should summarize the technical aspects rather than expound at length, and it should use the contributors as reservoirs of clinical experience. ///

Preconception and Preimplantation Diagnosis of Human Genetic Disease, Robert G. Edwards, editor, Cambridge University Press, New York, 1993, \$99.95.

Lisa Baumbach is assistant professor in the genetics division/department of pediatrics, Mailman Center for Child Development, P.O. Box 016820/D-280, Miami, FL 33101.