The third edition is considerably larger than its predecessor: 119 pages, 230 newly listed agents, references to new papers (into 1979), and additions of references to the older literature. The computerized format does not produce a stylish book, but the advantages of ease and rapidity of bringing the book up to date far outweigh aesthetics. Several technical changes, such as simplifying the citation numbers at the top of the pages, make this edition easier to use than the second edition.

The catalog should find a place next to the telephone of the clinical teratologist who receives frequent inquiries about exposures of pregnant women to a bewildering variety of agents. Although the information is not always the definitive word on the subject, it is information that is quickly accessible. Combined with experience and judgment the catalog helps the clinical teratologist render prompt and reasonably accurate risk estimates.

Considering the amount of information packed between its covers and the currency of that information, the catalog is a bargain at $25. In the previous two editions readers were provided with a form to return to Tom Shepard with criticisms, corrections (there are still a number of typos), and additions for future editions. The form has been dropped from the third edition but the wish to receive these comments is still stated in the introduction.

The first edition of the catalog was a noble achievement and the subsequent editions have each embodied major improvements. Tom Shepard deserves our support and help in continuing to update, improve, and expand hoped-for future editions.

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In 1970, Aubrey Milunsky first provided a comprehensive review of prenatal diagnosis, a three-part article in the New England Journal of Medicine. Then prenatal diagnosis was a novel investigative procedure, and now it is a safe clinical procedure used by many practitioners. Then there was an enormous amount of data produced by a few laboratories, and now there is a flood of information from many laboratories. It hardly seems possible to write a definitive reference as this tremendous data flux continues. However, this volume is intended to be the major reference for prenatal diagnosis of human disease. Prenatologists will turn to this book to answer questions which arise in the management of problems. Questions involving prenatal diagnosis are usually urgent, and decisions are usually irrevocable. We need to look at this book as a tool as important as any the physician may have.

Some of the most useful features of this volume are the summarizing tables. These are compilations of published studies and a worldwide survey which Milunsky conducted. This survey of 110 centers summarized the experience with 32,000 cases with amniocentesis. The tables have the advantage of extensive bibliographic data but the disadvantage of uneven collection and insufficient data, making conclusions impossible. For instance, one of the most difficult problems is that of chromosomal mosaicism in the amniotic fluid cells. It is often impossible to ascertain whether the mosaicism arose in the fetus, the amnion, or the culture. Table XII lists 82 instances of mosaicism in each. The text describes an instance in which repeat amniocentesis did not detect the mosaicism found at the first amniocentesis and later confirmed in the newborn. It is not possible to determine from the table how often mosaicism is confirmed on a subsequent culture. These data are all important for a working prenatologist.

The rapid pace of prenatal diagnosis has prevented the incorporation of the latest data into the body of the chapters. This has resulted in addenda at the end of each chapter. For instance, the Report of the British Medical Research Council was published in 1978 and reported a fetal loss of 1.5% due to amniocentesis. This report is described in detail in an addendum to Chapter 2. In this form, it is difficult for a reader to compare the lower incidences of previous studies with those of Britain. Perhaps this chapter should have been rewritten to achieve the needed clarity and perspective. Twelve chapters contain addenda.

This book is quite comprehensive and includes valuable chapters on some of the newer aspects of prenatal diagnosis such as “Diagnosis of Fetal Abnormalities by Ultrasound” by Stuart Campbell and “Radiographic Fetal Diagnosis” by N. Thorne Griscone. The chapter “Fetoscopy and Blood Sampling” is
especially helpful, since it relates the considerable experience of its authors—Mahoney and Hobbins. The technique of fetal blood sampling is described, and photographs of the vessels are given. The usefulness of this procedure to diagnose Factor VIII deficiency and the perils of using fetal blood samples to diagnose muscular dystrophy are described. This chapter also demonstrates the difficulty in making diagnoses of morphological abnormalities. The pictures of what the fetoscope operator sees would not encourage many persons to try their hand.

Prenatal diagnosis provides very good prospective genetic data. Populations at risk are selected, and the outcome is well documented. The maternal age effect in Down syndrome, the relationship of balanced chromosome translocations to malformation incidence, and the recurrence risks of chromosomal aneuploidy are all important data that will be obtained by the careful collection of experience with prenatal diagnosis. In Chapter 8 Milunsky reviews the risk figures for neural tube defects published in the literature and his considerable experience with the measurements of alphafetoprotein in amniotic fluid. He has performed almost 12,000 alphafetoprotein analyses and presents a very helpful prospective study. Of 1,717 cases studied because the parents had a previous child with a neural tube defect, only 1.5% had a recurrence. This is considerably below the 5–6% recurrence risk most often cited in the literature.

This chapter on neural tube defects is not so helpful for questions about the indications for amniocentesis. Milunsky states that there is a "similar consensus" on the need for amniocentesis both in women who have had a child with a neural tube defect and their sisters. He also states that "all couples at risk have a right to know their particular risks and to elect or reject the option of prenatal diagnosis." Since the risk for a recurrence of a NTD was 1.5% and presumably that for a third degree relative would be only 1/10 of that, it does not seem reasonable to believe that all couples have an equal right to obtain an amniocentesis. In areas where medical resources are severely taxed, some decisions not to perform amniocentesis will be made by health providers. Prenatal diagnosis is a discipline with a large share of ethical issues. The question of the right to a diagnostic test when facilities are limited is certainly one which will require further discussion.

Some of the less noteworthy chapters of the book include the use of trophoblasts for early prenatal diagnosis and the chapter on fetal cells in maternal circulation. Both subjects show some possibility for future study but have not proven helpful for prenatal diagnosis. The chapter on the role of infectious agents in birth defects is not helpful to the practicing perinatologist. The questions asked by patients are not addressed. For instance, most parents want to know the best way to determine if the fetus is affected after a maternal rubella exposure. They also want to know if a maternal vaccination for rubella will cause a birth defect. There is a great need to use amniocentesis to predict the risk of viral disease in the fetus. This chapter would be more helpful if it focused on questions a perinatologist would face rather than the general questions of the relationship of birth defects to maternal infection.

The largest chapter is that on the prenatal diagnosis of biochemical disorders. This chapter will be very helpful since a practitioner will be able to find the reference for almost all biochemical diagnoses which have been made. Presumably these references could be used to contact the laboratory and obtain direct help. In this regard, the bibliography could have been more selective—there are more than 1500 references in this chapter alone. Also Milunsky has failed to indicate the problem areas as clearly as he does the areas of success. It is not always evident that some tests, such as for cystic fibrosis, are still experimental.

As research moves ahead rapidly, the published results of that research will be immediately used for practical prenatal diagnosis. The startling impact of DNA restriction mapping is immediately apparent. It is imperative that publishing techniques be developed that foster the beneficial relationship between publication and practice. Data needs to be collected in a uniform and accurate manner so it can be used rapidly by publishers and editors. This book is important and could develop into a most essential tool, available to geneticists, in the same way as the McKusick Catalog. This book clearly illustrates the need for publishers to modernize and use the methods for data processing to speed the dissemination of important information.

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