Book Reviews

SERVICE AND EDUCATION IN MEDICAL GENETICS

This collection of papers is derived from the proceedings of the 7th Annual New York State Health Department Birth Defects Symposium. The theme is service and education in medical genetics. Included are articles from a related meeting of the American Society of Human Genetics in San Diego sponsored by the March of Dimes Birth Defects Foundation. The subject matter is divided into four sections: 1) epidemiological analyses, policy considerations, and economics; 2) genetic counseling and other genetic services in the community; 3) attitudes toward genetic counseling; and 4) education for medical genetics.

The prosaic title of this collection of papers in no way prepares the reader for the fascinating material included. Because of the inevitable lag time in publication, certain sections of the text are dated (e.g., discussions of prenatal detection of sickle cell disease make no reference to new techniques utilizing reduction endonucleases). In addition, some of the papers are superficial and introduce no new ideas. Nevertheless, many of the papers provide fascinating insight into issues of great importance in the future practice of the medical geneticist. This is not a book which will assist the geneticist in the care of the individual patient. Rather, its purpose is to survey the fundamental problems facing human geneticists in the 1980's. Some of the problems addressed are: 1) development of regional screening programs for prenatal detection of cytogenetic abnormalities and neural tube defects; what is the current state of organization of such programs? are they cost effective? how might such programs be organized? 2) third party payers in delivery of genetic services; what is their current role? how might this role be enhanced? 3) center-satellite systems for provision of genetic services; how are they organized and do they work? 4) genetic knowledge in high school students; are these young people being prepared to receive reproductive information based on current and evolving knowledge? 5) the impact of masters level genetic counseling training programs; what are graduates of these programs doing now and what is their future? 6) genetic knowledge of pediatricians and obstetricians in private practice; what do they know and how do they use it? 7) future educational needs in genetics; how should the public and the medical profession be educated to fulfill future needs?

Although the geneticist may not choose to invest $24.00 in a personal copy of this book, (s)he should certainly consider seriously the investment of the evening or two necessary to read it in its entirety. This comprehensive and fascinating collection can help us to face and answer some of the compelling clinical questions of the coming decade.

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This is a new edition of a book that is familiar to many teratologists and should be familiar to all. It consists of a computerized printout of an alphabetically arranged list of teratogenic agents. Each entry has a succinct account of reported studies and principal literature references. The list of agents is not confined to drugs and chemicals but also includes entries such as delayed fertilization, diabetes mellitus, emotional stress, and immobilization. The subject index lists agents by generic and brand names, which makes it quite easy to locate the desired information.

The catalog is valuable for the researcher who wants to know in brief what agents have been tested, how, and with what results. To be sure, the listing of agents and references is not exhaustive, but that is probably an impossible goal in an age in which chemicals and papers appear on the scene with overwhelming rapidity. The catalog does provide a very useful survey of a large number of agents and a large literature, and it is a place to start a more extensive literature search as needed. In the preface to this edition, Tom Shepard gives the reader the information needed to initiate more
in-depth searches, such as through ETIC/TOXLINE.

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 of 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